VOLUME 30 · AUTHOR INDEX

ABBS S *et al.* Discrepancy in the estimation of relative male and female mutation rates for Duchenne muscular dystrophy, 333 *abs*

ADAM S *et al.* Five year study of prenatal testing for Huntington's disease: demand, attitudes, and psychological assessment, 549

ADAMSON DJA *et al.* Molecular aspects of P53 in lymphoma and leukaemogenesis, 339 *abs*

ADÈS LC *et al.* Congenital heart malformation in Yunis-Varón syndrome: *brief paper*, 788
Hydrocephalus in Hajdu-Cheney syndrome: *letter*, 175

ADJIMAN M *see* BIENVENU T *et al*

AFFARA NA *see* RICHARDS FM *et al*
YATES JRW *et al*

AINSWORTH PJ *see* RODENHISER DI *et al*

AITKEN DA *et al.* Biochemical screening for chromosome abnormalities and neural tube defects in the first trimester, 336 *abs*

AKIYAMA J *see* PASSOS-BUENO MR *et al*

AL-AWADI SA *see* FARAG TI *et al*

AL GAZALI LI, QUAIFE R. Two sibs with unbalanced translocations in the Waardenburg gene region: *short report*, 607

AL-GAZALI LI *et al.* A syndrome of insulin resistance resembling leprechaunism in five sibs of consanguineous parents, 470

AL-JADER LN *et al.* Severity of chest disease in CF patients in relation to their genotypes: *letter*, 709

AL-OTHMAN SA *see* FARAG TI *et al*

ALDRED MA *et al.* Genetic localisation of RP2 type of X linked retinitis pigmentosa, 345 *abs*

ALDRED MJ *et al.* Identification of a nonsense mutation in the amelogenin gene (AMELX) in a family with X linked amelogenesis imperfecta (AIH1), 345 *abs*

ALSULIMAN IS *see* FARAG TI *et al*

ALTHOUSE R *see* CUCKLE HS *et al*

ALVIR J *see* MACDONALD ME *et al*

AMBERGER JS *see* MCKUSICK VA AND AMBERGER JS

AMBROSE CM *see* MACDONALD ME *et al*

ANDERSON MA *see* MACDONALD ME *et al*

ANDERSSON R *see* DRUGGE U *et al*

ANDREW SE *see* GOLDBERG YP *et al*
KREMER B *et al*

ANDRIA G *see* SPERLI D *et al*

ARIELI S *see* SHABTAI F *et al*

ARIMANY JL *see* SERES-SANTAMARIA A *et al*

ARNGRIMSSON R *et al.* Dyskeratosis congenita: three additional families show linkage to a locus in Xq28: *short report*, 618

AROSEMENA G *see* WARD J *et al*

ARROYO I *see* MARTINEZ-FRIAS M-L *et al*

ARTIFONI L *see* ROBINSON WP *et al*

ASHERSON P *see* OWEN MJ *et al*

AUDREZET MP *see* LISSENS W *et al*

AVIV H *see* ROBINSON WP *et al*

AXELROD FB *see* BLUMENFELD A *et al*

AYTER S *see* ÖZGÜÇ M *et al*

AZULAY J-P *see* YATES JRW *et al*

BACCICHETTI C *see* ROBINSON WP *et al*

BACSKULIN A *see* MEINDL A *et al*

BAKKER E *see* DE ROOIJ KE *et al*
NICHOLSON LVB *et al*

BALL D *see* SHAIKH S *et al*

BALL SP, JOHNSON KJ. The genetics of malignant hyperthermia: *review article*, 89

BARAITSER M *et al.* Cerebellar ataxia and ectodermal dysplasia in brothers: *short report*, 515
see also PATTON MA and BARAITSER M; REARDON W *et al*

BARANOV VS. Molecular diagnosis of some common genetic diseases in Russia and the former USSR: present and future: *medical genetics around the world* 141
see also IVASCHENKO T and BARANOV VS

BARBATO C *see* SPERLI D *et al*

BARBER JCK *see* FLYNN GA *et al*

BARLOW KM *see* BONTHRON DT *et al*

BARNES G *see* MACDONALD ME *et al*

BARNICOAT AJ *et al.* Two cases of aneuploidy and the fragile X syndrome, 338 *abs*

BARR DGD *see* BONTHRON DT *et al*

BARRATT A *see* MORRIS JA *et al*

BARRON LH *et al.* A study of the Huntington's disease associated trinucleotide repeat in the Scottish population, 1003
see also Simpson SA *et al*

BARTH ML *see* WALLEY AJ *et al*

BARTON DE *see* MAHER ER *et al*
ZHANG Y *et al*

BASHAWRI L *see* KNOX-MACAULAY HHM *et al*

BASTAKI L *see* FARAG TI *et al*

BATCH JA *et al.* Mutations of the androgen receptor gene identified in perineal hypospadias, 198

BATTIN J *see* LACOMBE D *et al*

BECKMANN JS *see* PASSOS-BUENO MR *et al*
PERAL B *et al*

BEGGS AH *see* MICHELS VV *et al*

BEIGHTON P *see* WALLIS GA *et al*

BELDJORD C *see* BIENVENU T *et al*

BELFROID RDM *see* DE ROOIJ KE *et al*

BELL C *et al.* Mutation detection in rhodopsin and peripherin genes in the Scottish retinitis pigmentosa population, 347 *abs*

BEN-SASSON E *see* SHABTAI F *et al*

BENJAMIN CM *et al.* Neurofibromatosis type 1 (NF1): knowledge, experience, and reproductive decisions of affected patients and families, 567

BENNETT CP *et al.* Deletion 9p and sex reversal: *short report*, 518

BENSAHEL H *see* MAROTEAUX P *et al*

BENSON MD *et al.* A transthyretin variant (alanine 49) associated with familial amyloidotic polyneuropathy in a French family, 117
A transthyretin variant (alanine 71) associated with familial amyloidotic polyneuropathy in a French family, 120
see also BENSON MD *et al*; ZELDENRUST SR *et al*

BENTLEY D *see* HOLDING C *et al*

BERMEJO E *see* MARTNEZ-FRIAS M-L *et al*

BERNARDI F *see* ROSSI E *et al*

BERNASCONI F *see* ROBINSON WP *et al*

BERNSTEIN J *see* KLEIMAN S *et al*

BERRY E *et al.* Biochemical screening for chromosome abnormalities: five years' experience of routine screening in the west of Scotland, 339 *abs*

BERRY P *see* MÜLLER U *et al*

BERTHIER M *see* BONNEAU D *et al*
VERLOES A *et al*

BERTIN TK *see* NORTHRUP H *et al*

BESNER A *see* MACKENZIE A *et al*

BEYER C *see* JENDERNY J *et al*

BHATTACHARYA SS *see* CURTIS A *et al*

BIEGEL JA *see* GOLDMUNTZ E *et al*

BIENVENU T *et al.* Male infertility as the only presenting sign of cystic fibrosis when homozygous for the mild mutation R117H: *letter*, 797
Severe cystic fibrosis in a child homozygous for the G542 nonsense mutation in the CFTR gene: *letter*, 621

BIESECKER LG *see* WANG C *et al*

BIRD E *see* MACDONALD ME *et al*

BISHOP DT *see* MUELLER RF AND BISHOP DT
BLIGHT A *see* MOSS C *et al*
BLOCH M *see* ADAM S *et al*
BLUMENFELD A *et al*. Exclusion of familial dysautonomia from more than 60% of the genome, 47
BOBROW M *see* HOLDING C *et al*
LANDELS EC *et al*
BOCHDALOFSKY V *see* THIES U *et al*
BOCIAN E *et al*. Triple structural mosaicism of chromosome 18 in a child with MR/MCA syndrome and abnormal skin pigmentation: *short report*, 614
BOCKEL B *see* THIES U *et al*
BOLUND L *see* TOMMERUP N *et al*
BONDUELLE M *see* LISSENS W *et al*
BONILLA E *see* MACDONALD ME *et al*
BONNEAU D *et al*. Usher syndrome type I associated with bronchiectasis and immotile nasal cilia in two brothers: *short report*, 253
X linked spastic paraplegia (SPG2): clinical heterogeneity at a single gene locus, 381
see also VERLOES A *et al*
BONTHRON DT *et al*. Parental consanguinity in the blepharophimosis, heart defect, hypothyroidism, mental retardation syndrome (Young-Simpson syndrome): *short report*, 255
BOOHENE J *see* CURTIS A *et al*
BOOK REVIEWS
BAUMANN N, FEDERICO A, SUZUKI K, *editors*. Late onset neurometabolic genetic disorders, 1992, 176
BEARN AG. Archibald Garrod – and the individuality of man, 1993, 973
BMA. Our genetic future: the science and ethics of genetic technology, 1992, 536
BONNE-TAMIR B, ADAM A, *editors*. Genetic diversity among Jews: diseases and markers at the DNA level, 1992, 352
BREUNING MH, DEVOTO M, ROMEO G, *editors*. Polycystic kidney disease. Contributions to nephrology vol 97, 1992, 87
BROCK DJH. Molecular genetics for the clinician, 1993, 712
BROCK DJH, RODECK CH, FERGUSON-SMITH MA, *editors*, Prenatal diagnosis and screening, 1992, 710
BUNDEY S. Genetics and neurology, 2nd edition, 1992, 710
CASSIDY SB, *editor*. Prader Willi syndrome and other chromosome 15q deletion disorders. NATO ASI Series H Cell Biology volume 61, 1992, 350
COHNE J, MALTER HE, TALANSKY B, GRIFO J. Micromanipulation of human gametes and embryos, 1992, 351
COLLINS MKL, *editor*. Practical molecular virology. Methods in Molecular Biology volume 8, 1991, 536
Cystic fibrosis and DNA tests: implications of carrier screening, 1992, 887
DAVIES KE. Molecular basis of inherited disease, 2nd edition, 1992, 447
DAVIES KE, TILGHMAN SM, *editors*. Genome analysis: genes and phenotypes, volume 3, 1992, 263
DRONAMRAJU KR, *editor*. The history and development of human genetics, 1992, 712
EDWARDS RG, *editor*. Fetal tissue transplants in medicine, 1992, 447
ELWOOD JM, LITTLE J, ELWOOD JH. Epidemiology and control of neural tube defects, 1993, 888
EVANS MI, *editor*. Reproductive risks and prenatal diagnosis, 1992, 712
EVERS-KIEBOOMS G, FRYNS JP, CASSIMAN JJ, VAN DEN BERGHE H, *editors*. Psychosocial aspects of genetic counselling, 1992, 448
FEDOROFF N, BOTSTEIN D, *editors*. The dynamic genome – Barbara McClintock's ideas in the century of genetics, 1993, 887
FRIEDMAN JM, DILL FJ, HAYDEN MR, MCGILLIVRAY BC. Genetics, 1992, 350
FRIEDMAN T, *editor*. Molecular genetic medicine, volume 2, 1992, 263
GARDNER KD, *editor*. The cystic kidney, 1992, 87
GROSVELD F, KOLLIAS G, *editors*. Transgenic animals, 1992, 623
HARDING AE, *editor*. Inherited ataxias. Advances in neurology. Volume 61, 1993, 711
KAUFMAN MH. The atlas of mouse development, 1992, 350
KEVLES DJ, HOOD L, *editors*. The code of codes. Scientific and social issues in the human genome project, 1992, 350
LUSIS AJ, ROTTER JI, SPARKES RS, *editors*. Molecular genetics of coronary artery disease: candidate genes and processes in atherosclerosis, 1992, 448
MCGUFFIN P, MURRAY R, *editors*. The new genetics of mental illness, 1991, 623
MCKUSICK VA. Mendelian inheritance in man, 10th edition, 2 volumes, 1992, 352
MILUNSKY A, *editor*. Genetic disorders and the fetus. Diagnosis, prevention and treatment, 3rd edition, 1992, 351
MILUNSKY A. Heredity and your family's health, 1993, 1043
MODELL B, KULIEV AM, WAGNER M. Community genetics services in Europe. Report on WHO survey, 1992, 264
NEALE MC, CARDON LR. Methodology for genetic studies of twins and families, 1992, 800
RICCARDI VM. Neurofibromatosis: phenotype, natural history and pathogenesis, 2nd edition, 1992, 351
ROBERTS DF, FUJIKI N, TORIZUKA K, *editors*. Isolation, migration and health, 1992, 888
ROONEY DE, CZEULKOWSKI BH, *editors*. Human cytogenetics: a practical approach, 1992, 710
SILVESTRONI IB. Microcitemie e anemia Mediterranea, 1992, 176
SOLOMON E, RAWLINGS C, *editors*. Human gene mapping 11. The eleventh international workshop on human gene mapping, 1992, 87
TSUJI K, AIZAWA M, SASAZUKI T, *editors*. HLA 1991, proceedings of the XI International Histocompatibility Workshop and Conference, volumes 1 and 2, 1992, 799
WEAVER DD. Catalog of prenatally diagnosed conditions, 2nd edition, 1992, 800
WIEDEMANN HR, KUNZE J, GROSSE FR, DIBBERN H. An atlas of clinical syndromes: a visual aid to diagnosis, 2nd edition, 1992, 711
WILLIAMS J, CECCARELLI A, SPURR N. Genetic engineering. Medical Perspectives Series, 1993, 711
BOOTH F *see* HILDES E *et al*
BORKOWSKA J *see* YATES JRW *et al*
BORSKI K *see* JARUZELSKA J *et al*
BOULOUX P *see* MEINDL A *et al*
BRAND N *see* KLEIMAN S *et al*
BRANDT CA *see* TOMMERUP N *et al*
BREAKEFIELD XO *see* BLUMENFELD A *et al*
BRIGGS MD *et al*. Molecular and genetic mechanisms in recurrent severe osteogenesis imperfecta, 339 *abs*
BRISCIOLI V *et al*. Trisomy 10qter confirmed by in situ hybridisation: *short report*, 601
BROCK D. Approaches to prenatal cystic fibrosis carrier screening: *letter*, 621
BROCK DJH *see* BARRON LH *et al*
MENNIE ME *et al*
SHRIMPTON AE *et al*
BRODERICK NJ *see* YOUNG ID *et al*
BROOK-CARTER PT *see* NELLIST M *et al*
BROOK JD *see* SHAW DJ *et al*
BROUX O *see* PASSOS-BUENO MR *et al*
BROWN GK *see* BROWN RM AND BROWN GK
BROWN JD *see* GOLDBERG YP *et al*
BROWN MM *see* HERNANDEZ D *et al*
BROWN RM, BROWN GK. X chromosome inactivation and the diagnosis of X linked disease in females, 177
BROWN WT *see* OOSTRA BA *et al*
BRÜCKL W *see* MEINDL A *et al*
BRZUSTOWICZ L *see* MACDONALD ME *et al*
BUCKLE VJ *see* FLYNN GA *et al*
BUDARF ML *see* DRISCOLL DA *et al*
GOLDMUNTZ E *et al*

BULAWA E *see* BOCIAN E *et al*
BULLMAN H *see* JACOBS PA *et al*
BULTEAU C *see* BONNEAU D *et al*
BUNDEY S, YOUNG ID. Low segregation ratios in autosomal recessive disorders: *annotation*, 449
see also WATKISS E *et al*
BUNDEY S *et al*. A prospective study on the effect of inbreeding: 1986–1992, 334 *abs*
BUNTINCX IM *et al*. Syndromic association of cleft palate, bilateral choanal atresia, curly hair, and congenital hypothyroidism: *short report*, 427
BURN J *et al*. Conotruncal anomaly face syndrome is associated with a deletion within chromosome 22q11, 822
see also WADEY R *et al*; WILSON DI *et al*
BURNELL LD *see* EVANS DGR *et al*
BURNETT JC *see* MICHELS VV *et al*
BURT AM *see* BONTHRON DT *et al*
BUSHBY KMD *et al*. Variability of clinical, genetic and protein abnormalities in manifesting carriers of Xp21 muscular dystrophy, 337 *abs*
see also NICHOLSON LVB *et al*
BUTLER MG *see* DE VRIES BBA *et al*
BUTLER TJ *see* NICHOLSON LVB *et al*
BUTT AM *et al*. Probable de novo 17q duplication (q11.2–q21.1): a newly recognised chromosomal syndrome in a child with Klinefelter's syndrome: *short report*, 436
BUXTON J *see* CHAN C-TJ *et al*
BYE MR *see* SCHULMAN M *et al*
BYERS PH *see* WALLIS GA *et al*

CABROL S *see* SCHNEID H *et al*
CALABRO A *see* LUNGAROTTI MS AND CALABRO A
CALIEBE A *see* JENDERNY J *et al*
CALLEN DF *et al*. High resolution mapping of interstitial long arm deletions of chromosome 16: relationship to phenotype, 828
CALZOLARI E *et al*. The Emilia Romagna birth defects register: goals and accomplishments, 344 *abs*
CAMASCHELLA C *see* DIANZANI I *et al*
CAMERA G *et al*. Limb/pelvis-hypoplasia/aplasia syndrome (Al-Awadi/Raas-Rothschild syndrome): report of two Italian sibs and further confirmation of autosomal recessive inheritance, 65
CAMERON A *see* HILDES E *et al*
CAMPANELLA G *see* DI MAIO L *et al*
CANZIANI F *see* DE VRIES BBA *et al*
CARNEVALE A *see* OROZCO L *et al*
CARTER NP *see* ZHENG Y *et al*
CARVALHO AA *see* PASSOS-BUENO MR *et al*
CASALI M *see* ROSSI E *et al*
CASSIMAN J-J *see* DECORTE R AND CASSIMAN J-J
CASTLE D *see* SHAIKH S *et al*
CATANZARITE V *see* WEYERTS LK *et al*
CAVANAGH N *see* REARDON W *et al*
CHA J-H *see* MACDONALD ME *et al*
CHAN C-TJ *et al*. Molecular mechanisms in Angelman syndrome: a survey of 93 patients, 895
CHANCELLOR A *see* KING A *et al*
CHARLES SJ *see* MAHER ER *et al*
ZHANG Y *et al*
CHATKUPT S *et al*. Waardenburg syndrome and myelomeningocele in a family: *short report*, 83
see also CHATKUPT S *et al*
CHAUDHARY R *see* CLARKSON PA *et al*
CHAUDHARY S *see* SHAW DJ *et al*
CHÁVEZ M *see* OROZCO L *et al*
CHENG X-J *see* CHAN C-TJ *et al*
CHENG XJ *see* CLAYTON-SMITH J *et al*
CHIAFFONI GP *see* TURCO AE *et al*
CHINOY NJ *see* RADHAKRISHNA U *et al*
CHITTY L *et al*. Fetal echogenic bowel and cystic fibrosis in low risk fetuses, 333 *abs*
CHITTY LS *see* SELLER MJ *et al*
CHIUMELLO G *see* ROSSI E *et al*
CHIURAZZI P *see* KOZK L *et al*
CHIZARI F *see* DRUGGE U *et al*
CHOGNOT D *see* STOLL C *et al*
CHOW G *see* FARRELL SA *et al*
CHRISTIE BS *see* WHITE JM *et al*
CHRISTODOULOU Z *see* HIRST MC *et al*
CHU C *et al*. Screening for occult Y mosaicism in girls with Turner's syndrome, 346 *abs*
CHUNG E *et al*. Linkage analysis of infantile pyloric stenosis and markers from chromosome 9q11–q33: no evidence for a major gene in this candidate region, 393
CICHON S *see* NÖTHEN MM ET AL
CLARK C *et al*. Early experience of fragile X mutation detection in the north-east of Scotland, 346 *abs*
CLARKE AJ *see* PARSONS EP AND CLARKE AJ
CLARKE DJ *see* PRASHER VP *et al*
CLARKSON PA *et al*. Mutational screening of the Wilms's tumour gene, WT1, in males with genital abnormalities, 767
CLAVEY V *see* HUMPHRIES S *et al*
CLAYTON-SMITH J *et al*. Duplication of chromosome 15 in the region 15q11–13 in a patient with developmental delay and ataxia with similarities to Angelman syndrome: *short report*, 529
see also CHAN C-TJ *et al*
CLOW CL *see* MITCHELL J *et al*
COCHRANE S *et al*. A linkage study of X linked Charcot-Marie-Tooth disease (CMTX) using highly informative loci, 345 *abs*
COCKBURN D *et al*. A strategy for the investigation of Duchenne and Becker muscular dystrophies in the diagnostic genetics laboratory based on accurate carrier detection, 347 *abs*
COCKWELL AE *see* DENNIS NR *et al*
COFFEY R *see* CHUNG E *et al*
COHEN D *see* PASSOS-BUENO MR *et al*
COLE T *et al*. A search for uniparental disomy in Sotos syndrome, 342 *abs*
COLE WG *et al*. The clinical features of spondyloepiphyseal dysplasia congenita resulting from the substitution of glycine 997 by serine in the alpha-1(II) chain of type II collagen, 27
COLEMAN MP *see* GLASS IA *et al*
COLEMAN R *et al*. Interaction of incontinentia pigmenti and factor VIII mutations in a female with biased X inactivation, resulting in haemophilia, 497
COLLEY A *see* BENJAMIN CM *et al*
COLLIER D *see* SHAIKH S *et al*
COLLINS AL *et al*. Holoprosencephaly: a family showing dominant inheritance and variable expression, 36
see also DENNIS NR *et al*
COLMAN SM *see* ZHENG Y *et al*
COMPTON ME *see* MENNIE ME *et al*
CONCOLINO D *see* SPERLI D *et al*
CONDE JB *see* MACDONALD ME *et al*
CONNEALLY PM *see* DI MAIO L *et al*
MACDONALD ME *et al*
CONNOR JM *et al*. A three generation family with fibrodysplasia ossificans progressiva: *brief paper*, 687
see also ARNGRIMSSON R *et al*; NELLIST M *et al*
CONSALEZ GG *et al*. Cloning of a candidate gene for Menkes syndrome spanning the MNK translocation breakpoint, 334 *abs*
CONTE A *see* VALERIO D *et al*
COOKE A *et al*. Molecular genetic studies of Angelman syndrome and Prader-Willi syndrome patients, 348 *abs*
COOKSON H *see* WILLIAMS C *et al*
COOMBS RC *see* POWELL CVE *et al*
COONAR AS *et al*. Two cases of 5q deletions in subjects with familial adenomatous polyposis: possible link with Caroli's disease and Beale's syndrome, 340 *abs*
see also HODGSON SV *et al*
COOPER PJ *et al*. A balanced whole arm reciprocal translocation resulting in three different adverse pregnancy outcomes: *short report*, 417
CORBETT JA *see* PRASHER VP *et al*
COSKUN T *see* ÖZGÜÇ M *et al*
COSTA FF *see* MARTINS CSB *et al*
COSTA T *see* DIXON JW *et al*
COTTON RGH *see* DIANZANI I *et al*
RAMUS SJ *et al*

COTTRELL S *et al.* Detection of mutations in part of the APC gene in FAP families and sporadic colorectal cancer, 339 *abs*
see also HODGSON SV *et al*
COULTER-MACKIE MB *see* RODENHISER DI *et al*
COWELL JK *see* MORRIS JA *et al*
COX TM *see* MISTRY PK AND COX TM
CRADDOCK N *see* SHAIKH S *et al*
CRAGG H *see* WILLIAMSON M *et al*
CRAUFURD D, DODGE A. Mutation size and age at onset in Huntington's disease, 1008
CROCQ M-A *see* OWEN MJ *et al*
CROLLA JA *see* COOPER PJ *et al*
DENNIS NR *et al*
CROQUETTE M-F *see* OUDET C *et al*
CROSS GS, SHARIF AL. Mutation analysis of fragile X families, 346 *abs*
CROSS I *see* BURN J *et al*
CROSS S *see* DEAN JCS *et al*
CROSSEY PA *see* RICHARDS FM *et al*
CROW S *see* SHAW DJ *et al*
CUCKLE HS *et al.* Sex differences in the location of a spina bifida lesion: *letter*, 262
CUPPLES LA *see* VAN DUIJN CM *et al*
CURNOW RN *et al.* Relative risk and risky relatives, 344 *abs*
CURTIS A *et al.* Absence of cystic fibrosis mutations in a large Asian population sample and occurrence of a homozygous S549N mutation in an inbred Pakistani family: *short report*, 164
see also NICHOLSON LVB *et al*
CZEIZEL AE *et al.* An epidemiological study of isolated split hand/foot in Hungary, 1975–1984, 593

DAAR S *see* WHITE JM *et al*
DAB I *see* LISSENS W *et al*
DAHL SP *et al.* Genetic heterogeneity of Usher syndrome type II, 843
DALLAPICCOLA B *see* GERBONI S *et al*
DALTON JD *see* GORDON PL *et al*
DANESINO C *see* ROSSI E *et al*
DANIELSSON M *see* DRUGGE U *et al*
DAVID TJ *see* POWELL CVE *et al*
DAVIDSON HR. A large family with patent ductus arteriosus and unusual face, 503
DAVIDSON HR *et al.* An unusual cystic fibrosis family, 343 *abs*
DAVIDSON MJ *see* SIMPSON SA *et al*
DAVIDSON R *see* BARRON LH *et al*
SHRIMPTON AE *et al*
DAVIES HR *see* CLARKSON PA *et al*
DAVIES JP *et al.* Sequence variations in the first exon of alpha-galactosidase A, 658
DAVIES KE *see* FLYNN GA *et al*
GLASS IA *et al*
HIRST MC *et al*
KNOX-MACAULAY HHM *et al*
SUTHERS GK *et al*
DAVIES SJ, HUGHES HE. Imprinting in Albright's hereditary osteodystrophy, 101
DAVISON EV *see* MOSS C *et al*
DAW S *see* WADEY R *et al*
DE ALMEIDA JCC *et al.* Another example favouring the location of BPES at 3q2: *letter*, 86
DE BELLEROCHE J *see* KING A *et al*
DE DIE-SMULDERS C *see* SMITS APT *et al*
DE DIE-SMULDERS CEM *et al.* Severe intrauterine growth retardation, blepharophimosis, and cylindrical nose with midline groove: a new syndrome? *short report*, 525
DE DINECHIN O *et al.* Workshop of the Commission of the European Communities on Ethics of Human Genome Analysis: survey of the European discussion: *conference report*, 257
DE KONING GANS PAM *see* DE ROOIJ KE *et al*
DE LA CHAPELLE A. Disease gene mapping in isolated human populations: the example of Finland: *medical genetics around the world*, 857
see also HÄSTBACKA J *et al*
DE MIRANDA A *see* LLERENA JC *et al*
DE ROOIJ KE *et al.* Dynamic mutation in Dutch Huntington's disease patients: increased paternal repeat instability extending to within the normal size range, 996
DE SILVA D *et al.* Congenital skull defect and recurrent abdominal desmoids: a variant of familial adenomatous polyposis? 342 *abs*
DE SMET NMAFA *see* SCHRANDER-STUMPEL CTRM *et al*
DE VRIES BBA *et al.* Clinical and molecular studies in fragile X patients with a Prader-Willi-like phenotype, 761
DE YOUNG M *see* MACDONALD ME *et al*
DEAN J *see* MIEDZYBRODZKA Z *et al*
DEAN JCS *et al.* Counselling pitfalls in Romano-Ward syndrome: *letter*, 886
Evidence of genetic and phenotypic heterogeneity in the Romano-Ward syndrome, 947
Wolf-Hirschhorn syndrome: paternal origin of a submicroscopic deletion of chromosome 4, 348 *abs*
see also MIEDZYBRODZKA ZH *et al*; PACKE GE *et al*
DEAN M *see* WILL K *et al*
DECORTE R, CASSIMAN J-J. Forensic medicine and the polymerase chain reaction technique: *review article*, 625
DECRUYENAERE M *et al.* Perception of predictive testing for Huntington's disease by young women: preferring uncertainty to certainty? 557
DEGET F *see* ZERRES K *et al*
DEGRAVE W *see* LLERENA JC *et al*
DELATTRE S *see* HUMPHRIES S *et al*
DELEZOIDE A-L *see* MORICHON-DELVALLEZ N *et al*
DEMUTH S *see* LENK U *et al*
DEN DUNNEN JT *see* DE ROOIJ KE *et al*
NICHOLSON LVB *et al*
DENNIS NR *et al.* Three patients with ring (X) chromosomes and a severe phenotype, 482
see also COLLINS AL *et al*; JACOBS PA *et al*
DESAGER K *see* BUNTINCX IM *et al*
DESMYTTERE S *see* LISSENS W *et al*
DEVADAS K *see* AL-GAZALI LI *et al*
DEYMEER F *see* YATES JRW *et al*
DI MAIO L *et al.* Onset symptoms in 510 patients with Huntington's disease, 289
Suicide risk in Huntington's disease, 293
DIANZANI I *et al.* Molecular analysis of contiguous exons of the phenylalanine hydroxylase gene: identification of a new PKU mutation, 228
Two new mutations in the dihydropteridine reductase gene in patients with tetrahydrobiopterin deficiency, 465
DIFFER A-M *et al.* Analysis of the unstable CTG repeat in myotonic dystrophy, 341 *abs*
DIXON JW *et al.* Mosaicism for duplication 12q (12q13–q24.2) in a dysmorphic male infant: *short report*, 70
DOCHERTY Z *see* BENNETT CP *et al*
DODGE A *see* CRAUFURD D AND DODGE A
DOKAL I *see* ARNGRIMSSON R *et al*
DOLK H, SELLER MJ. Neural tube defects: a survey of lesion descriptions made by different European pathologists, 942
DONNAI D *see* BENJAMIN CM *et al*
JOUET M *et al*
LANDY SJ AND DONNAI D
DRABKIN HA *see* PERICAK-VANCE MA *et al*
DREESEN JCFM *see* SMITS APT *et al*
DRISCOLL D *see* GOLDMUNTZ E *et al*
DRISCOLL DA *et al.* Prevalence of 22q11 microdeletions in DiGeorge and velocardiofacial syndromes: implications for genetic counselling and prenatal diagnosis, 813
DRISCOLL DJ *see* MICHELS VV *et al*
DROOG RP *see* DE DIE-SMULDERS CEM *et al*
DRUGGE U *et al.* Familial amyloidotic polyneuropathy in Sweden: a pedigree analysis, 388
DRY KL *et al.* Characterisation of a 340 kb YAC and cDNA enrichment from a region spanning the locus for retinitis pigmentosa (*RP3*), 335 *abs*
DUNBAR H *see* SELLER MJ *et al*
DUNDAR M *et al.* Screening for the G985 medium chain

acyl-CoA dehydrogenase deficiency mutation in Scottish women, 346 *abs*
DURBACH N, HAYDEN MR. George Huntington, the man behind the eponym: *portraits in medical genetics*, 406
DURE L *see* MACDONALD ME *et al*
DUYAO MP *see* MACDONALD ME *et al*

EDWARDS TJC *et al*. Humeroradioulnar synostosis in a patient with lambdoid synostosis: *short report*, 81
EGGERS S *et al*. Facioscapulohumeral muscular dystrophy: aspects of genetic counselling, acceptance of preclinical diagnosis, and fitness, 589
ELBORN S *see* AL-JADER LN *et al*
ELITZUR A *see* KLEIMAN S *et al*
ELLES RG *et al*. Diagnosis of APKD by genetic markers and ultrasonography, 332 *abs*
ELLIOTT J *et al*. A case of deletion 14(q22.1→q22.3) associated wtih anophthalmia and pituitary abnormalities: *short report*, 251
ELLIS I *see* WALLEY AJ *et al*
ELLIS IH *see* LANDELS EC *et al*
ELSAWI MM *et al*. Male fertility in Noonan syndrome, 342 *abs*
EMANUEL BS *see* DRISCOLL DA *et al*
EMANUEL BS *see* GOLDMUNTZ E *et al*
EMERY AEH, EMERY MLH. Edward Meryon (1809–1880) and muscular dystrophy: *portraits in medical genetics*, 506
EMERY MLH *see* EMERY AEH AND EMERY MLH
ENBLAD P *see* RICHARDS FM *et al*
ERDEM H *see* ÖZGÜÇ M *et al*
ESSIG Y-P *see* KOUSSEFF BG *et al*
EUROPEAN COMMUNITY HUNTINGTON'S DISEASE COLLABORATIVE STUDY GROUP. Ethical and social issues in presymptomatic testing for Huntington's disease: a European Community collaborative study, 1028
EVANS BAJ *see* BATCH JA *et al*
EVANS DGR *et al*. Complications of the naevoid basal cell carcinoma syndrome: results of a population based study, 460
Gorlin syndrome and congenital anomalies in medulloblastoma, 337 *abs*
EVANS K *et al*. Cone-rod dystrophy associated with subretinal neovascularisation and gingival dystrophy, 342 *abs*
EVERS-KIEBOOMS G *see* DECRUYENAERE M *et al*
EYRE H *see* CALLEN DF *et al*

FARAG TI *et al*. The newly recognised limb/pelvis-hypoplasia/aplasia syndrome: report of a Bedouin patient and review, 62
FARLOW M *see* ZELDENRUST SR *et al*
FARNDON PA *see* EVANS DGR *et al*
GLASS IA *et al*
MOSS C *et al*
FARRELL SA *et al*. Interstitial deletion of chromosome 10q23: a new case and review: *short report*, 248
FARRER LA *see* VAN DUIJN CM *et al*
FEIGHERY C *see* PADAYACHEE M *et al*
FELDMAN E *see* JOUET M *et al*
FENSOM AH *see* LANDELS EC *et al*
WALLEY AJ *et al*
FEREC C *see* LISSENS W *et al*
FERGUSON C *see* RICHARDS A *et al*
FERGUSON-SMITH MA *see* MAHER ER *et al*
RICHARDS FM *et al*
YATES JRW *et al*
ZHENG Y *et al*
FERRAIOLO G *see* CAMERA G *et al*
FERRERO GB *see* DIANZANI I *et al*
FIELDER A *see* GLASS IA *et al*
FILIPOWICZ J *see* JARUZELSKA J *et al*
FIMMERS R *see* NÖTHEN MM ET AL
FINN A *see* PADAYACHEE M *et al*
FISHER AM *see* DENNIS NR *et al*
FLANNERY AV *see* FLYNN GA *et al*
FLETCHER NA, FOLEY J. Parental age, genetic mutation, and cerebral palsy, 44
FLINTER FA *see* BUTT AM *et al*
FLORIDIA G *see* BRISCIOLI V *et al*
ROSSI E *et al*
FLYNN GA *et al*. Identification of the *FRAXE* fragile site in two families ascertained for X linked mental retardation, 97
FOLEY J *see* FLETCHER NA AND FOLEY J
FONKNECHTEN N *see* BIENVENU T *et al*
FORREST SM *see* RAMUS SJ *et al*
FOUGEROUSSE F *see* PASSOS-BUENO MR *et al*
FRANZONI E *see* ROBINSON WP *et al*
FRASER FC *see* GARABEDIAN BH AND FRASER FC
FREISINGER P *see* MAROTEAUX P *et al*
FRIEND JAR *see* MIEDZYBRODZKA ZH *et al*
FRUCHART J-C *see* HUMPHRIES S *et al*
FRYBURG JS, GREER KE. Epidermal naevi and bullous aplasia cutis congenita in a neonate: *brief paper*, 962
FRYNS J-P, MOERMAN P. Short limbed dwarfism, genital hypoplasia, sparse hair, and vertebral anomalies: a variant of Ellis-van Creveld syndrome: *short report*, 322
FRYNS J-P *et al*. Profound mental retardation, characteristic facies with midfacial hypoplasia and premature frontotemporal balding, muscular hypotrophy, and small patellae in two unrelated male patients: *short report*, 319
see also DE DIE-SMULDERS CEM *et al*; DE VRIES BBA *et al*; HIRST MC *et al*; SCHRANDER-STUMPEL CTRM *et al*; SOEKARMAN D AND FRYNS JP
FULLWOOD P *see* GLASS IA *et al*
FURMAN JMR *see* DAHL SP *et al*

GAMMARO L *see* TURCO AE *et al*
GARABEDIAN BH, FRASER FC. Upper and lower neural tube defects: an alternate hypothesis, 849
GARDINER RM *see* CHUNG E *et al*
GARDNER-MEDWIN D *see* HARMON DL *et al*
NICHOLSON LVB *et al*
SMITH RA and
GARDNER-MEDWIN D
GARRETT C *see* COLLINS AL *et al*
GAUNT ML *see* LYNCH SA *et al*
GAYTHER S *et al*. Germline rearrangement at the *APC* locus detected by PFGE, 340 *abs*
GENET SA *see* COLEMAN R *et al*
GENTIL V *see* ZATZ M *et al*
GENUARDI M *see* KOZÁK L *et al*
GERBONI S *et al*. Coarctation of the aorta, interrupted aortic arch, and hypoplastic left heart syndrome in three generations: *short report*, 328
GERDES A *et al*. Congenital myopathy with fibre type disproportion: a family with a chromosomal translocation t(10;17) may indicate candidate gene regions, 348 *abs*
GEUTJENS J *see* FRYNS J-P *et al*
GHETTI B *see* ZELDENRUST SR *et al*
GIL R *see* BONNEAU D *et al*
GILES MG *see* GLASS IA *et al*
GILFILLAN A *see* MENNIE ME *et al*
GILL M *see* SHAIKH S *et al*
ZATZ M *et al*
GIMELLI G *et al*. ICF syndrome with variable expression in sibs: *short report*, 429
GINJAAR IB *see* NICHOLSON LVB *et al*
GLASS IA *et al*. Ear anomalies, clefting, and limb reduction defects: a new autosomal recessive condition? 342 *abs*
Genetic mapping of a cone and rod dysfunction (Åland Island eye disease) to the proximal short arm of the human X chromosome, 1044
GLOVER TW *see* WANG C *et al*
GOERTZ K *see* HOLMES GE *et al*
GOLDBERG YP *et al*. Familial predisposition to recurrent mutations causing Huntington's disease: genetic risk to sibs of sporadic cases, 987
see also KREMER B *et al*
GOLDMAN ASH, HULTÉN MA. Chiasma frequency, quadrivalent orientation, and first meiotic segregation in a human male reciprocal translocation

heterozygote t(15;20)(q13;q13.1) analysed by dual colour fluorescence in situ hybridisation, 336 *abs*
GOLDMUNTZ E *et al.* Microdeletions of chromosomal region 22q11 in patients with congenital conotruncal cardiac defects, 807
GOMEZ F *see* MACDONALD ME *et al*
GONÇALVES MS *see* MARTINS CSB *et al*
GONZALEZ MT *see* SCHULMAN M *et al*
GOOD P *see* GLASS IA *et al*
GOODBURN SF *et al.* Maternal serum screening for Down's syndrome in East Anglia, 338 *abs*
GOODCHILD MC *see* AL-JADER LN *et al*
GOODEVE JA *see* BUTT AM *et al*
GOODSHIP J *see* BURN J *et al*
WADEY R *et al*
WILSON DI *et al*
GORDON AG. Aphasia, deafness, or mental retardation: *letter*, 262
GORDON PL *et al.* Elucidation of the centromere involvement in an inversion (13) by fluorescent in situ hybridisation: *short report*, 414
GORIN MB *see* DAHL SP *et al*
GORSKI JL *see* MCDONALD MT AND GORSKI JL
GOSCH A, PANKAU R. Chromosome abnormalities and Williams-Beuren syndrome: *letter*, 886
GOURMELEN M *see* SCHNEID H *et al*
GOVAERTS LCP *see* SMITS APT *et al*
GRADE K *see* LENK U *et al*
GRAHAM CA *see* MORRISON PJ *et al*
GRANT D *see* BENNETT CP *et al*
GRANT J *see* WILLIAMSON M *et al*
GRAY J *see* MACDONALD ME *et al*
GREAVES MF *see* ZHENG Y *et al*
GREEN J *et al.* Psychosocial issues raised by a familial ovarian cancer register, 575
see also RICHARDS FM *et al*
GREEN JS *see* ZHANG Y *et al*
GREEN PM *see* LANDELS EC *et al*
GREENBERG CR *see* HILDES E *et al*
GREENBERG F. DiGeorge syndrome: an historical review of clinical and cytogenetic features: *review article*, 803
GREER KE *see* FRYBURG JS AND GREER KE
GREWAL PK *see* HIRST MC *et al*
GRIFFIN DK *et al.* Preimplantation diagnosis of sex in X linked disease by fluorescent in situ hybridisation, 332 *abs*
GRINBLAT J *see* SHABTAI F *et al*
GROTE W *see* JENDERNY J *et al*
GRUNDBACHER FJ. The contribution of genetic factors to the pathogenesis of type I (insulin dependent) diabetes mellitus: *letter*, 533
GRUNEWALD I *see* LENK U *et al*
GUDNASON V *see* HUMPHRIES S *et al*
GUIDI O *see* VERLOES A *et al*
GUILHERME L *see* PASSOS-BUENO MR *et al*
GUSELLA JF *see* BLUMENFELD A *et al*
MACDONALD ME *et al*
GUSTAVSON KH *see* RICHARDS FM *et al*

HAAN EA *see* ADÈS LC *et al*
CALLEN DF *et al*
EDWARDS TJC *et al*
HACKETT GA *see* ZHENG Y *et al*
HÅGÅ P *see* ÖRSTAVIK KH *et al*
HAINES JL *see* BLUMENFELD A *et al*
HAITES N *see* MIEDZYBRODZKA Z *et al*
HAITES NE *see* MIEDZYBRODZKA ZH *et al*
HAJIANPOUR AK *et al.* The dilemma of two cell populations in Fanconi anaemia, 340 *abs*
HALB A *see* STOLL C *et al*
HALFORD S *see* WADEY R *et al*
HALL JG. CATCH 22: *editorial*, 801
see also SCHRANDER-STUMPEL CTRM *et al*
HALL RK *see* COLE WG *et al*
HALLEY DJJ *see* DE VRIES BBA *et al*
HAMILTON TJ *see* MOTUM PI *et al*
HANAUER A *see* OUDET C *et al*
HANDYSIDE AH. Preimplantation diagnosis of cystic fibrosis by DNA amplification from single cells, 332 *abs*
HANKE R *see* LENK U *et al*
HANSMANN I *see* CALLEN DF *et al*
HANSON PJV *see* HODGSON SV *et al*
HARCOURT SA *see* LEHMANN AR *et al*
HARDING AE *see* REIK W *et al*
SIMPSON SA *et al*
HARDY C *et al.* Flanking markers for the gene for the Gorlin syndrome, 345 *abs*
HARDY J *see* KING A *et al*
HARLEY HG *see* SHAW DJ *et al*
HARMON DL *et al.* Two new mutations in a late infantile Tay-Sachs patient are both in exon 1 of the beta-hexosaminidase alpha subunit gene, 123
HARPER JI *see* COLEMAN R *et al*
HARPER PS. A specific mutation for Huntington's disease: *editorial*, 975
Journal of Medical Genetics – Cytogenetics editor: *editor's note*, 1043
Psychosocial genetics: an emerging scientific discipline: *editorial*, 537
see also MACMILLAN JC *et al*; SHAW DJ *et al*
HARRIS A *see* WALLEY AJ *et al*
HARRIS JB *see* NICHOLSON LVB *et al*
HARRIS R, RHIND JA. Medical genetics in the reformed NHS and in Europe, 334 *abs*
HARRIS R, RHIND JA. The specialty of clinical genetics: European Society of Human Genetics survey, 147
see also BENJAMIN CM *et al*; DE DINECHIN O *et al*
HARVEY I *see* SHICKLE D AND HARVEY I
HÄSTBACKA J *et al.* Prenatal diagnosis of diastrophic dysplasia with polymorphic DNA markers, 265
HAUSMANOWA-PETRUSEWICZ I *see* YATES JRW *et al*
HAWKINS JR *see* BENNETT CP *et al*
HAWLEY PR *see* HODGSON SV *et al*
HAYDEN MR *see* ADAM S *et al*
DURBACH N AND HAYDEN MR
GOLDBERG YP *et al*
KREMER B *et al*
HELLEBOSTAD M *see* ÖRSTAVIK KH *et al*
HENDRIKS RW *see* PADAYACHEE M *et al*
HERNÁNDEZ C *see* PERAL B *et al*
HERNANDEZ D *et al.* A family showing no evidence of linkage between the ataxia telangiectasia gene and chromosome 11q22–23, 135
HERRMANN FH *see* SCHRÖDER W ET AL
HEWETT DR *et al.* The screening of the fibrillin gene FBN1 for causal mutations in Marfan syndrome, 348 *abs*
HIGGS DR *see* WHITE JM *et al*
HILDES E *et al.* Impact of genetic counselling after neonatal screening for Duchenne muscular dystrophy, 670
HIRSH A *see* WILLIAMS C *et al*
HIRST MC *et al.* Origins of the fragile X syndrome mutation, 647
see also FLYNN GA *et al*
HODGSON SV *et al.* Two cases of 5q deletions in patients with familial adenomatous polyposis: possible link with Caroli's disease, 369
HOFMAN A *see* VAN DUIJN CM *et al*
HOGG C *see* PATTON MA *et al*
HOLDER SE, WINTER RM. Otopalatodigital syndrome type II: *syndrome of the month*, 310
HOLDER SE *et al.* Velocardiofacial syndrome in a mother and daughter: variability of the clinical phenotype, 825
see also VINTINER GM *et al*
HOLDING C *et al.* Development and validation of laboratory procedures for preimplantation diagnosis of Duchenne muscular dystrophy, 903
HOLLINGSWORTH Z *see* MACDONALD ME *et al*
HOLLOWAY S *see* BARRON LH *et al*
HOLMES GE *et al.* Continuation of a case report: *letter*, 445
HOLMGREN G *see* DRUGGE U *et al*
HOMFRAY T *et al.* The Martin Bell family revisited, 341 *abs*
HOPWOOD JJ *see* SCHRÖDER W *et al*
HORI T *see* SUZUMORI K *et al*
HORN N *see* TOMMERUP N *et al*

HOSENFELD D *see* MEINDL A *et al*
HOULDEN H *see* KING A *et al*
HÖWELER CJ *see* SCHRANDER-STUMPEL CTRM *et al*
HOWELLS DW *see* DIANZANI I *et al*
HUANG S-Z *see* ZENG Y-T *et al*
HUANG Y *see* ZENG Y-T *et al*
HUERTAS H *see* MARTÍNEZ-FRIAS M-L *et al*
HUGGINS M *see* ADAM S *et al*
HUGHES HE *see* DAVIES SJ AND HUGHES HE
REARDON W *et al*
HUGHES IA *see* BATCH JA *et al*
CLARKSON PA *et al*
HUMPHREY IJ *see* EDWARDS TJC *et al*
HUMPHRIES S *et al*. Six DNA polymorphisms in the low density lipoprotein receptor gene: their genetic relationship and an example of their use for identifying affected relatives of patients with familial hypercholesterolaemia, 273
HUNT N *see* SHAIKH S *et al*
HUSON SM *et al*. The Oxford neurofibromatosis clinic: the first two years' experience, 337 *abs*
see also SUTHERS GK *et al*

IVASCHENKO T, BARANOV VS. Rapid and efficient PCR/*Sty*I test for identification of common mutation R408W in phenylketonuria patients: *technical note*, 153

JACKSON A *see* CURTIS A *et al*
JACKY PB *see* OOSTRA BA *et al*
JACOBS HK *see* HILDES E *et al*
JACOBS PA *et al*. Population studies of the fragile X: a molecular approach, 454
see also DENNIS NR *et al*
JAMES WH. The combinations of the sexes of familial cases of neural tube defect: *letter*, 447
JARDINE PE *et al*. Absence of facial involvement may differentiate a scapular onset form of muscular dystrophy from FSH dystrophy, 338 *abs*
The scleroatrophic syndrome of Huriez, 342 *abs*
JARUZELSKA J *et al*. Genetic background of clinical homogeneity of phenylketonuria in Poland, 232
see also WITT M *et al*
JÄRVELÄ IE *et al*. 46,XX/69, XXX diploid-triploid mixoploidy with hypothyroidism and precocious puberty: *brief paper*, 966
JEFFERY S *see* PATTON MA *et al*
JENDERNY J *et al*. Transmission of a ring chromosome 18 from a mother with 46,XX/47,XX, +r(18) mosaicism to her daughter, resulting in a 46,XX, r(18) karyotype: *brief paper*, 964
see also MEINDL A *et al*
JENKINS B *see* MACDONALD ME *et al*
JENKINS D *et al*. Hypomelanosis of Ito associated with mosaicism for trisomy 7 and apparent 'pseudomosaicism' at amniocentesis: *brief paper*, 783
JENNINGS K *see* DEAN JCS *et al*
JENTSCH TJ *see* KOCH MC *et al*
JOHNSON KJ *see* BALL SP AND JOHNSON KJ
JOHNSON MA *see* NICHOLSON LVB *et al*
JOHNSON P *see* NELLIST M *et al*
JOHNSON WG *see* CHATKUPT S *et al*
JONES A *see* SWEATMAN A *et al*
JONES MC *see* WEYERTS LK *et al*
JONES MH *see* MAHER ER *et al*
JONES T *see* HODGSON SV *et al*
JOUET M *et al*. Refining the genetic location of the gene for X linked hydrochephalus within Xq28, 214
JULIEN J *see* BENSON MD *et al*
JUNG JH *see* RODENHISER DI *et al*
JUNG M *see* DE ALMEIDA JCC *et al*

KABACK MM *see* LANDELS EC *et al*
KAITILA I *see* HÄSTBACKA J *et al*
KALIL J *see* PASSOS-BUENO MR *et al*
KAMATH S *see* HOLDER SE *et al*
KAMPER J *see* TOMMERUP N *et al*
KAPLAN F *see* MITCHELL J *et al*
KAPLAN J *see* BONNEAU D *et al*
OUDET C *et al*
KAPLAN JC *see* BIENVENU T *et al*
KEARNEY A *see* MOTUM PI *et al*
KELLY KF *see* MIEDZYBRODZKA ZH *et al*
KENWRICK S *see* JOUET M *et al*
KERZIN-STORRAR L *see* BENJAMIN CM *et al*
KETTNER M *see* DE DINECHIN O *et al*
KHALIL M *see* AL-GAZALI LI *et al*
KILPATRICK M *see* GLASS IA *et al*
KIMBERLING WJ *see* DAHL SP *et al*
KING A *et al*. Absence of linkage between chromosome 21 loci and familial amyotrophic lateral sclerosis: *short report*, 318
KING-UNDERWOOD L *see* HUMPHRIES S *et al*
KINGSTON H *see* BENJAMIN CM *et al*
KINNON C *see* PADAYACHEE M *et al*
SWEATMAN A *et al*
KLEIMAN S *et al*. Phenylketonuria: variable phenotypic outcomes of the R261Q mutation and maternal PKU in the offspring of a healthy homozygote, 284
KLOSSEK J-M *see* BONNEAU D *et al*
KNIGHT LA *et al*. Extra G positive band on the long arm of chromosome 9: *short report*, 613
KNIGHT SJL *see* FLYNN GA *et al*
HIRST MC *et al*
KNOBLOCH O *et al*. Direct versus indirect molecular diagnosis of fragile X mental retardation in 40 German families at risk, 193
KNOX-MACAULAY HHM *et al*. X linked recessive thrombocytopenia: *short report*, 968
KOCH DJ *see* MAHER ER *et al*
KOCH L *see* DE DINECHIN O *et al*
KOCH MC *et al*. Evidence for genetic homogeneity in autosomal recessive generalised myotonia (Becker), 914
KODAJ I *see* CZEIZEL AE *et al*
KONDO I *see* SUZUMORI K *et al*
KOROSHETZ W *see* MACDONALD ME *et al*
KOULISCHER L *see* VERLOES A *et al*
KOUSSEFF BG *et al*. Complex chromosome rearrangement with ankyloblepharon filiforme adnatum: *short report*, 167
KOZK L *et al*. Mapping of a gene for non-specific X linked mental retardation: evidence for linkage to chromosomal region Xp21.1–Xp22.3: *brief paper*, 866
KRÄFT U *see* LENK U *et al*
KREMER B *et al*. Molecular analysis of late onset Huntington's disease, 991
see also GOLDBERG YP *et al*
KREMER C *see* BONNEAU D *et al*
KRETZ K *see* WILLIAMSON M *et al*
KRISHNAN VHR *see* PRASHER VP *et al*
KUCZORA I *see* WITT M *et al*
KUNKEL LM *see* MICHELS VV *et al*
KUPKE KG *see* MÜLLER U *et al*
KWIATKOWSKI DJ *see* NELLIST M *et al*

LACOMBE D *et al*. Split hand/split foot deformity and LADD syndrome in a family: overlap between the EEC and LADD syndromes: *brief paper*, 700
LADUSANS EJ *see* EVANS DGR *et al*
LALATTA F *see* BRISCIOLI V *et al*
LANDELS EC *et al*. Further investigation of the HEXA gene intron 9 donor splice site mutation frequently found in non-Jewish Tay-Sachs disease patients from the British Isles, 479
LANDY SJ, DONNAI D. Incontinentia pigmenti (Bloch-Sulzberger syndrome): *syndrome of the month*, 53
LANDY SJ *et al*. A clinical and genetic study of incontinentia pigmenti, 341 *abs*
LANE R *see* KING A *et al*
LANE S *see* CALLEN DF *et al*
LANGSLET A *see* ÖRSTAVIK KH *et al*
LARKINS S *see* MOSS C *et al*
LATHROP GM *see* PERAL B *et al*
LATIF F *see* RICHARDS FM *et al*
LATOS-BIELENSKA A *see* WITT M *et al*
LAUNDON C *et al*. Turner's syndrome detection by MSAFP/Free beta (hCG) screening, 340 *abs*

LAURENCE KM. Trends in the number of births with spina bifida, 338 *abs*
LAURILA P *see* HÄSTBACKA J *et al*
LAVORGNA F *see* VALERIO D *et al*
LE BOUC Y *see* SCHNEID H *et al*
LE MERRER M *see* BONNEAU D *et al*
MAROTEAUX P *et al*
LECHEVALIER B *see* BENSON MD *et al*
LEE D *see* WARD J *et al*
LEE N *see* PATTON MA *et al*
LEHMANN AR *et al*. Cockayne's syndrome: correlation of clinical features with cellular sensitivity of RNA synthesis to UV irradiation, 679
LEIGH DA *see* NEILAN BA *et al*
LENK U *et al*. Alternative splicing of dystrophin mRNA complicates carrier determination: report of a DMD family, 206
Non-isotopic analysis of single strand conformation polymorphism (SSCP) in the exon 13 region of the human dystrophin gene, 951
LENOIR G *see* BIENVENU T *et al*
LENZ W *see* CZEIZEL AE *et al*
LEO D *see* CAMERA G *et al*
LERMAN M *see* RICHARDS FM *et al*
LERONE M *see* GIMELLI G *et al*
LEVINSKY R *see* SWEATMAN A *et al*
LEVINSKY RJ *see* PADAYACHEE M *et al*
LEVY N *see* LANDELS EC *et al*
LEWIS N *see* REARDON W *et al*
LEWIS RA *see* NORTHRUP H *et al*
LEZANA JL *see* OROZCO L *et al*
LI Y *see* WANG F AND LI Y
LIEBAERS I *see* LISSENS W *et al*
LIEBERT CB *see* BLUMENFELD A *et al*
LIEPNIEKS J *see* BENSON MD *et al*
LIM-STEELE J *see* LANDELS EC *et al*
LINDSAY S *see* GLASS IA *et al*
LINEHAN WM *see* RICHARDS FM *et al*
LINFORTH GH, PLAHA DS. Predictive testing for Huntington's disease: experience with the multiallelic probe pKP1.65, 347 *abs*
LINFORTH GH *et al*. Predictive testing in a one generation Huntington's disease family, 347 *abs*
LISSENS W *et al*. Mild pulmonary, but severe hepatic disease in a cystic fibrosis patient homozygous for a frameshift mutation in the regulatory domain of the CFTR: *letter*, 446
LISTON WA *see* MENNIE ME *et al*
LLERENA JC *et al*. Cystic fibrosis and deafness: *letter*, 621
see also DE ALMEIDA JCC *et al*
LLOYD J *et al*. A T^{+6} to C^{+6} mutation in the donor splice site of COL3A1 IVS7 causes exon skipping and results in Ehlers-Danlos syndrome type IV, 376
see also RICHARDS A *et al*
LO KK *see* VINTINER GM *et al*
LO Y-MD *et al*. Prenatal sex determination from maternal peripheral blood using the polymerase chain reaction, 336 *abs*
LOEB DB *see* PERICAK-VANCE MA *et al*
LOPEZ M *see* WITT M *et al*
LORENZ C *see* KOCH MC *et al*
LOSEKOOT M *see* DE ROOIJ KE *et al*
LOUGHLIN JA *et al*. Molecular analysis of the allelic expression of the cartilage specific type II collagen gene COL2A1 in osteoarthritis, 348 *abs*
LOVERING R *see* SWEATMAN A *et al*
LOWRY RB. Absent fibula and craniosynostosis: a 25 year follow up: *letter*, 445
LUCENTE DE *see* BLUMENFELD A *et al*
LUCKEL JC *see* STOLL C *et al*
LUCOTTE G *see* BENSON MD *et al*
LUNGAROTTI MS, CALABRO A. Weyer's ulnar ray/oligodactyly syndrome: *letter*, 533
LUNT PW *see* COLLINS AL *et al*
CONNOR JM *et al*
LUSH M *see* RICHARDS FM *et al*
LUZZATTO L *see* ARNGRIMSSON R *et al*
LYNCH SA *et al*. Campomelic dysplasia: evidence of autosomal dominant inheritance: *brief paper*, 683
LYONNET S *see* JARUZELSKA J *et al*
MAAYAN C *see* BLUMENFELD A *et al*
MCCONVILLE CM *see* HERNANDEZ D *et al*
MCDONALD BL *see* NEILAN BA *et al*
MCDONALD-MCGINN D *see* CALLEN DF *et al*
MCDONALD-MCGINN DM *see* DRISCOLL DA *et al*
GOLDMUNTZ E *et al*
MACDONALD ME *et al*. Gametic but not somatic instability of CAG repeat length in Huntington's disease, 982
MCDONALD MT, GORSKI JL. Nager acrofacial dysostosis: *syndrome of the month*, 779
MACDONALD N *see* SHRIMPTON AE *et al*
MACFARLANE H *see* MACDONALD ME *et al*
MCGUFFIN P *see* OWEN MJ *et al*
MACKAY K *et al*. An RT-PCR-SSCP strategy for detection of mutations in the gene encoding the alpha 1 chain of type I collagen: application to the analysis of four osteogenesis imperfecta patients, 335 *abs*
MACKENZIE A *et al*. Rapid diagnosis of infantile spinal muscular atrophy by direct amplification of amniocyte and CVS DNA: *short report*, 162
MACKENZIE MJ *et al*. Elucidation of possible mechanisms of sex chromosome non-disjunction using centromere map analysis, 335 *abs*
MCKEOWN C *see* PADAYACHEE M *et al*
MCKUSICK VA, AMBERGER JS. The morbid anatomy of the human genome: chromosomal location of mutations causing disease: *review article*, 1
MCMAHON R *see* ZHANG Y *et al*
MACMILLAN JC, QUINN NP. 15th International World Federation of Neurology Workshop on Huntington's Disease, 31 August–3 September 1993, Boston, Massachusetts, USA: *conference report*, 1039
MACMILLAN JC *et al*. Identification of an expanded CAG repeat in the Huntington's disease gene (IT15) in a family reported to have benign hereditary chorea, 1012
X linked adrenomyeloneuropathy misdiagnosed as multiple sclerosis in a manifesting heterozygote, 338 *abs*
MACPHERSON J *see* JACOBS PA *et al*
MACPHERSON JN *see* FLYNN GA *et al*
MCSHANE A *see* BARAITSER M *et al*
MAGALHÃES HA *see* MARTINS MC *et al*
MAGNANI I *see* ROSSI E *et al*
MAGUIRE S *see* AL-JADER LN *et al*
MAHER ER *et al*. Common mechanism of tumourigenesis in five types of von Hippel-Lindau disease neoplasms, 339 *abs*
Evaluation of molecular genetic diagnosis in the management of familial adenomatous polyposis coli: a population based study, 675
see also REIK W *et al*; RICHARDS FM *et al*
MALCOLM S *see* CHAN C-TJ *et al*
CLAYTON-SMITH J *et al*
DAVIES JP *et al*
PADAYACHEE M *et al*
VINTINER GM *et al*
MALISZEWSKA CT *see* PRASHER VP *et al*
MALTBY EL *see* ELLIOTT J *et al*
MANT R *see* OWEN MJ *et al*
MANT R *see* SHAIKH S *et al*
MARAFIE MJ *see* FARAG TI *et al*
MARCHAND D *see* LACOMBE D *et al*
MARCOLONGO A *see* TURCO AE *et al*
MARIE SKN *see* PASSOS-BUENO MR *et al*
MAROTEAUX P *et al*. Dominant carpotarsal osteochondromatosis: *brief paper*, 704
MARTENS PR *see* GORDON PL *et al*
MARTIN K *see* JENKINS D *et al*
MARTÍNEZ-FRÍAS M-L *et al*. Lethal short rib-polydactyly syndromes: further evidence for their overlapping in a continuous spectrum, 937
MARTINS CSB *et al*. Molecular characterisation of beta thalassaemia heterozygotes in Brazil: *letter*, 797
MARTINS MC *et al*. Hereditary anaemias in Portugal: epidemiology, public health significance, and control, 235
MARTINS RR *see* DE ALMEIDA JCC *et al*

MASCHIO G *see* TURCO AE *et al*
MATHEW C *see* HOLDING C *et al*
MATHEW CG *see* WALLIS GA *et al*
MATTIOLI L *see* HOLMES GE *et al*
MATUSZAK R *see* JARUZELSKA J *et al*
MAYALL ES *see* WILLIAMS C *et al*
MAYNARD S *see* WANG C *et al*
MAZURCZAK T *see* BOCIAN E *et al*
MÉHES K. Delayed speech development, facial asymmetry, strabismus, and transverse ear lobe creases: a new syndrome? *short report*, 76
MEHTA D *see* BUTT AM *et al*
MEINDL A *et al*. Analysis of a terminal Xp22.3 deletion in a patient with six monogenic disorders: implications for the mapping of X linked ocular albinism, 838
MEITINGER T *see* MEINDL A *et al*
MELO J *see* MARTINS MC *et al*
MELO MS *see* ZATZ M *et al*
MENDOZA A *see* WEYERTS LK *et al*
MENNIE ME *et al*. Prenatal screening for cystic fibrosis: psychological effects on carriers and their partners, 543
MERCIER B *see* LISSENS W *et al*
MEREDITH R *et al*. Audiometric identification of carriers of Usher's syndrome type II, 343 *abs*
MESCHINO W *see* ADAM S *et al*
METTEY R *see* BONNEAU D *et al*
MICHALCZAK K *see* WITT M *et al*
MICHELS VV *et al*. Dystrophin analysis in idiopathic dilated cardiomyopathy, 955
MIDDLETON-PRICE H *see* SWEATMAN A *et al*
MIEDZYBRODZKA Z *et al*. A new approach to prenatal cystic fibrosis carrier screening: *letter*, 86; *letter*, 621
Cystic fibrosis in Grampian: a population study, 343 *abs*
MIEDZYBRODZKA ZH *et al*. Antenatal carrier screening for cystic fibrosis: comparative evaluation of two approaches, 343 *abs*
Prevalence of cystic fibrosis mutations in the Grampian region of Scotland: *short report*, 316
see also PACKE GE *et al*
MILLER FA *see* MICHELS VV *et al*
MILNER BJ *et al*. p53 mutations in ovarian cancer, 339 *abs*
MILTON PJ *see* ZHENG Y *et al*
MINFORD AMB *see* LYNCH SA *et al*
MINGARELLI R *see* GERBONI S *et al*
MISTRY PK, COX TM. The glucocerebrosidase locus in Gaucher's disease: molecular analysis of a lysosomal enzyme: *review article*, 889
MITCHELL J *et al*. What young people think and do when the option for cystic fibrosis carrier testing is available, 538
MOERMAN P *see* FRYNS J-P AND MOERMAN P
MOGHRABI N *et al*. DNA markers for the analysis of the Crigler Najjar syndrome, type I, 346 *abs*
MOHAMMED FM *see* FARAG TI *et al*
MOHAMMED S *et al*. A variant of the Li-Fraumeni syndrome? 335 *abs*
MOLL PP *see* MICHELS VV *et al*
MÖLLER C *see* DAHL SP *et al*
MOMMA K *see* BURN J *et al*
MOORE AT *see* MAHER ER *et al*
ZHANG Y *et al*
MORELL R. Association between schizophrenia and homozygosity at the dopamine D3 receptor gene: *letter*, 708
MORENO F *see* PERAL B *et al*
MORENO M *see* OROZCO L *et al*
MORGAN G *see* SWEATMAN A *et al*
MORICHON-DELVALLEZ N *et al*. Holoprosencephaly and sacral agenesis in a fetus with a terminal deletion 7q36→7qter: *short report*, 521
MORRIS JA *et al*. Retinoblastoma: a possible link with low level radiation: *conference report*, 440
MORRIS LL *see* ADS LC *et al*
MORRISON PJ, NEVIN NC. Heterozygote frequency, mutation rate, fertility, and genetic fitness in the N Ireland Huntington's disease population, 344 *abs*
MORRISON PJ *et al*. DNA storage and duplicate sampling: lessons learnt from testing for Huntington's disease: *letter*, 1042
Ocular motor abnormalities in early Huntington's disease, 344 *abs*
Significant linkage disequilibrium between the Huntington's disease locus and markers at loci D4S10, D4S95, and D4S111 in Northern Ireland, 1018
see also MACMILLAN JC *et al*; REIK W *et al*
MOSCOWITZ C *see* MACDONALD ME *et al*
MOSS C *et al*. Epidermal mosaicism and Blaschko's lines, 752
MOTUM PI *et al*. Filipino β° thalassaemia: a high Hb A_2 β° thalassaemia resulting from a large deletion of the 5' β globin gene region, 240
MUELLER RF, BISHOP DT. Autozygosity mapping, complex consanguinity, and autosomal recessive disorders: *letter*, 798
MUELLER RF *et al*. Homozygosity mapping of autosomal recessive disorders and complex consanguinity, 345 *abs*
MÜLLER U *et al*. Second polar body incorporation into a blastomere results in 46,XX/69,XXX mixoploidy: *short report*, 597
MULTANI AS *see* RADHAKRISHNA U *et al*
MUÑIZ F *see* SERES-SANTAMARIA A *et al*
MUNNICH A *see* BONNEAU D *et al*
JARUZELSKA J *et al*
MURA M *see* ROSSI E *et al*
MURRELL J *see* ZELDENRUST SR *et al*
MURTHY DSK *see* FARAG TI *et al*
MURTON F *see* GREEN J *et al*
MYERS RH *see* MACDONALD ME *et al*

NAGASE H *see* MAHER ER *et al*
NAKAMURA Y *see* MAHER ER *et al*
NAM D *see* WHITE JM *et al*
NAPOLITANO G *see* DI MAIO L *et al*
NARCISI P *see* LLOYD J *et al*
RICHARDS A *et al*
NEILAN BA *et al*. The rapid analysis of dystrophin gene deletions shows variable electrophoretic mobility: *technical note*, 60
NELLIST M *et al*. Identification of markers flanking the tuberous sclerosis locus on chromosome 9 (*TSC1*), 224
NELSON DL *see* KNOBLOCH O *et al*
NELSON R *see* CURTIS A *et al*
NEMETH AH *see* GLASS IA *et al*
NERI G *see* KOZÁK L *et al*
NETO JBG *see* DE ALMEIDA JCC *et al*
NEVIN NC *see* MACMILLAN JC *et al*
MORRISON PJ *et al*
NEWBURY-ECOB R *et al*. Manifesting heterozygosity in Norrie's disease, 346 *abs*
NEWBURY-ECOB RA *et al*. Sex linked valvular dysplasia: *brief paper*, 873
NICHOLSON LVB *et al*. Integrated study of 100 patients with Xp21 linked muscular dystrophy using clinical, genetic, immunochemical, and histopathological data. Part 1. Trends across the clinical groups, 728: Part 2. Correlations within individual patients, 737: Part 3. Differential diagnosis and prognosis, 745
NIERMEIJER MF *see* DE VRIES BBA *et al*
NORBURY G *et al*. Analysis of the expansion associated with myotonic dystrophy in various tissues from fetuses predicted to be at high risk, 341 *abs*
NORRIS PG *see* LEHMANN AR *et al*
NORTHRUP H *et al*. Variability of expression in tuberous sclerosis, 41
NOTARANGELO LD *see* PADAYACHEE M *et al*
NÖTHEN MM *et al*. Association versus linkage studies in psychosis genetics: *annotation*, 634
Excess of homozygosity at the dopamine D3 receptor gene in schizophrenia not confirmed: *letter*, 708
NUNES KJ *see* PERICAK-VANCE MA *et al*

O'BRIEN J *see* WILLIAMSON M *et al*

OGATA T *et al.* Chromosomal localisation of a gene(s) for Turner stigmata on Yp, 918
OLEY CA *et al.* Terminal deletions of 2q: is there a consistent phenotype? 338 *abs*
OLIM G *see* MARTINS MC *et al*
OOSTRA B *see* RICHARDS FM *et al*
OOSTRA BA *et al.* Guidelines for the diagnosis of fragile X syndrome, 410
see also DE VRIES BBA *et al*; SMITS APT *et al*
OPPERMANN H-C *see* MEINDL A *et al*
ORIOT D *see* VERLOES A *et al*
OROZCO L *et al.* Frequency of ΔF508 in a Mexican sample of cystic fibrosis patients, 501
ØRSTAVIK KH *et al.* Possible X linked congenital mitochondrial cardiomyopathy in three families, 269
OTTO M *see* KOCH MC *et al*
OUDET C *et al.* Characterisation of a highly polymorphic microsatellite at the *DXS207* locus: confirmation of very close linkage to the retinoschisis disease gene, 300
OWEN G *see* AL-JADER LN *et al*
OWEN M *see* SHAIKH S *et al*
OWEN MJ, MCGUFFIN P. Association and linkage: complementary strategies for complex disorders: *comment*, 638
OWEN MJ *et al.* Association between schizophrenia and homozygosity at the dopamine D3 receptor gene. *letter*, 708
see also MCGUFFIN P and OWEN MJ
ÖZALP I *see* ÖZGÜÇ M *et al*
OZELIUS LJ *see* BLUMENFELD A *et al*
ÖZGÜÇ M *et al.* Mutation analysis in Turkish phenylketonuria, 129

PACKE GE *et al.* Severity of chest disease in CF patients in relation to their genotypes: *letter*, 709
PADAYACHEE M *et al.* Mapping of the X linked form of hyper IgM syndrome (HIGM1), 202
PADOVANI EM *see* TURCO AE *et al*
PANKAU R *see* GOSCH A AND PANKAU R
PAPENHAUSEN P *see* KOUSSEFF BG *et al*
PAPI L *see* ROSSI E *et al*
PARKER K *see* CHUNG E *et al*
PARSONS EP, CLARKE AJ. Genetic risk: women's understanding of carrier risks in Duchenne muscular dystrophy, 562
PASSOS-BUENO MR *et al.* Evidence of genetic heterogeneity in the autosomal recessive adult forms of limb-girdle muscular dystrophy following linkage analysis with 15q probes in Brazilian families, 385
see also EGGERS S *et al*; ZATZ M *et al*
PASTORES GM *see* MICHELS VV *et al*
PATERSON J *see* JOUET M *et al*
PATERSON JS, CONNOR JM. Fragile X syndrome families in the west of Scotland, 341 *abs*
PATTE F *see* BONNEAU D *et al*
PATTERSON MN *see* BATCH JA *et al*
CLARKSON PA *et al*
PATTON MA, BARAITSER M. Cutis laxa and the Costello syndrome: *letter*, 622
PATTON MA *et al.* Congenital nystagmus cosegregating with a balanced 7;15 translocation: *short report*, 526
PAYNE SJ *see* MAHER ER *et al*
PEARSON C *see* ADÈS LC *et al*
PEDERSEN S *see* TOMMERUP N *et al*
PEISSEL B *see* TURCO AE *et al*
PELZ F *see* KNOBLOCH O *et al*
PEMBREY ME *see* CHAN C-TJ *et al*
CHUNG E *et al*
CLAYTON-SMITH J *et al*
PENCHASZADEH G *see* MACDONALD ME *et al*
PENNEY J *see* MACDONALD ME *et al*
PERAL B *et al.* Estimating locus heterogeneity in autosomal dominant polycystic disease (ADPKD) in the Spanish population, 910
PERICAK-VANCE MA *et al.* Genetic mapping of dinucleotide repeat polymorphisms and von Hippel-Lindau disease on chromosome 3p25–26, 487
PETERLIN B *et al.* Prevalence and incidence of Duchenne (DMD) and Becker (BMD) muscular dystrophies in Slovenia, 344 *abs*
PETRUSCHKA L *see* SCHRÖDER W *et al*
PEZZOLO A *see* GIMELLI G *et al*
PFEIFFER RA *see* TRAUTMANN U *et al*
PHIPPS ME *see* RICHARDS FM *et al*
PIGNATTI PF *see* TURCO AE *et al*
PIKUS A *see* DAHL SP *et al*
PISTOIA V *see* GIMELLI G *et al*
PITT DB *see* RAMUS SJ *et al*
PLAHA DS, NEWBURY-ECOB RA. Different molecular mutations in male sibs with Duchenne muscular dystrophy, 347 *abs*
POMPONI G *see* KOZK L *et al*
PONZONE A *see* DIANZANI I *et al*
POPE FM *see* LLOYD J *et al*
RICHARDS A *et al*
PORTEOUS M *see* BARRON LH *et al*
PORTEOUS MEM *et al.* A new mental retardation syndrome mapping to the pericentromeric region of the X chromosome, 337 *abs*
POST JG *see* SMITS APT *et al*
POWELL CVE *et al.* Poland anomaly with contralateral ulnar ray defect: *short report*, 423
POZZOLO S *see* CAMERA G *et al*
PRASHER VP *et al.* Deletion of chromosome 2(p11–p13): case report and review: *short report*, 604
PRICE CM *see* ZHENG Y *et al*
PROPPING P *see* NÖTHEN MM *et al*
PULLEN I *see* MENNIE ME *et al*
PURVIS-SMITH S *see* OGATA T *et al*

QUAIFE R *see* AL GAZALI LI AND QUAIFE R
QUARRELL OWJ *see* MACMILLAN JC *et al*
QUINN NP *see* MACMILLAN JC AND QUINN NP

RADHAKRISHNA U *et al.* Polydactyly: a study of a five generation Indian family, 296
RAMALHO AS *see* MARTINS CSB *et al*
RAMANI P *see* BENNETT CP *et al*
RAMOS-ARROYO M *see* MACDONALD ME *et al*
RAMUS S *see* DIANZANI I *et al*
RAMUS SJ *et al.* Comparison of genotype and intellectual phenotype in untreated PKU patients, 401
RAVINE D *et al.* Phenotype and genotype heterogeneity in autosomal dominant polycystic kidney disease, 333 *abs*
RAYMOND F *see* BONNEAU D *et al*
READ AP *see* PADAYACHEE M *et al*
REARDON W *et al.* A new form of familial ataxia, deafness, and mental retardation: *brief paper*, 694
Consanguinity, cardiac arrest, hearing impairment, and ECG abnormalities: counselling pitfalls in the Romano-Ward syndrome: *short report*, 325
see also BARAITSER M *et al*
REEKERS ABA *see* SCHRANDER-STUMPEL CTRM *et al*
REIK W *et al.* Age at onset in Huntington's disease and methylation at *D4S95*, 185
REISS J *see* WILL K *et al*
REN Z-R *see* ZENG Y-T *et al*
REY F *see* JARUZELSKA J *et al*
REY J *see* JARUZELSKA J *et al*
REYNOLDS B *see* ELLIOTT J *et al*
RHIND JA *see* HARRIS R AND RHIND JA
RICHARD I *see* PASSOS-BUENO MR *et al*
RICHARDS A *et al.* The substitution of glycine 661 by arginine in type II collagen produces mutant molecules with different thermal stabilities and causes Ehlers-Danlos syndrome type IV: *brief paper*, 690
see also LLOYD J *et al*
RICHARDS FM *et al.* Detailed genetic mapping of the von Hippel-Lindau disease tumour suppressor gene, 104; correction, 528
RICHARDS RI *see* SUTHERLAND GR AND RICHARDS RI
RICHARDSON M *see* ADÈS LC *et al*
RICHARDSON RJ *see* CURTIS A *et al*
RICHARDSON W *see* HOLMES GE *et al*
RICKER K *see* KOCH MC *et al*
RIMMER S *see* EVANS DGR *et al*

RIMMLER JB *see* PERICAK-VANCE MA *et al*
ROBB SA *see* BENNETT CP *et al*
ROBERTS C *see* WADEY R *et al*
ROBERTS R *see* HOLDING C *et al*
ROBINSON N *et al*. Experience of DMD/BMD carrier detection by in situ hybridisation, 347 *abs*
ROBINSON WP *et al*. Uniparental disomy explains the occurrence of the Angelman or Prader-Willi syndrome in patients with an additional small inv dup(15) chromosome, 756
RODEHEFFER RJ *see* MICHELS VV *et al*
RODENHISER DI *et al*. A genetic study of neurofibromatosis type 1 (NF1) in south-western Ontario. II A PCR based approach to molecular and prenatal diagnosis using linkage, 363
RODRIGUES MO *see* MARTINS MC *et al*
ROGERS JG *see* COLE WG *et al*
ROMAN EO *see* OUDET C *et al*
ROONEY V *see* JACOBS PA *et al*
ROOS RAC *see* DE ROOIJ KE *et al*
ROSES AD *see* ZELDENRUST SR *et al*
ROSSETTI S *see* TURCO AE *et al*
ROSSI E *et al*. Types, stability, and phenotypic consequences of chromosome rearrangements leading to interstitial telomeric sequences, 926
see also BRISCIOLI V *et al*
ROUSSEAU F *see* OOSTRA BA *et al*
ROWICKA G *see* BOCIAN E *et al*
ROY N *see* MACKENZIE A *et al*
ROZET J-M *see* BONNEAU D *et al*
RUDNIK-SCHÖNEBORN S *see* ZERRES K *et al*
RUNDLE SA *see* SHAW DJ *et al*
RUSSELL G *see* MIEDZYBRODZKA ZH *et al*
RUTTER N *see* NEWBURY-ECOB RA *et al*
RYLEY HC *see* AL-JADER LN *et al*
RYSIECKI G *see* HERNANDEZ D *et al*

SABATINO G *see* GERBONI S *et al*
SAGLIO G *see* DIANZANI I *et al*
SALCEDO M *see* OROZCO L *et al*
SALEEBA JA *see* DIANZANI I *et al*
RAMUS SJ *et al*
SALO MK *see* JÄRVELÄ IE *et al*
SALONEN R *see* HÄSTBACKA J *et al*
SALONEN RK *see* JÄRVELÄ IE *et al*
SALVIN J *see* DRISCOLL DA *et al*
SAMPSON J *et al*. Early experience in preclinical diagnosis of familial adenomatous polyposis (FAP) in Wales, 340 *abs*
SAMPSON JR *see* NELLIST M *et al*
SAN MILLÁN JL *see* PERAL B *et al*
SANCHEZ-RAMOS J *see* MACDONALD ME *et al*
SANDER H *see* WOLFF G *et al*
SANDGREN O *see* DRUGGE U *et al*
SANTAVUORI P *see* JÄRVELÄ IE *et al*
SCALONA M *see* VALERIO D *et al*
SCAMBLER P *see* BURN J *et al*
WILSON DI *et al*
SCAMBLER PJ *see* HOLDER SE *et al*
WADEY R *et al*
SCHIMKE RN *see* HOLMES GE *et al*
SCHINZEL AA *see* ROBINSON WP *et al*
SCHLÖSSER M see Will K *et al*
SCHMIDTKE J *see* WILL K *et al*
SCHNEID H *et al*. Parental allele specific methylation of the human insulin-like growth factor II gene and Beckwith-Wiedemann syndrome, 353
SCHRANDER-STUMPEL CTRM *et al*. Arthrogryposis, ophthalmoplegia, and retinopathy: confirmation of a new type of arthrogryposis: *short report*, 78
SCHRÖDER W *et al*. Carrier detection of Hunter syndrome (MPS II) by biochemical and DNA techniques in families at risk, 210
SCHUFFENHAUER S *see* CALLEN DF *et al*
MEINDL A *et al*
SCHULMAN M *et al*. Airway abnormalities in Jarcho-Levin syndrome: a report of two cases, 875
SCHWAB SG *see* NÖTHEN MM *et al*
SCHWARTZ G *see* KLEIMAN S *et al*
SCRIVEN PN *see* HODGSON SV *et al*
SCRIVER CR *see* MITCHELL J *et al*
SEED M *see* HUMPHRIES S *et al*
SEGUES B *see* OUDET C *et al*
SEIDLITZ G *see* SCHRÖDER W *et al*
SEKI N *see* SUZUMORI K *et al*
SELICORNI A *see* BRISCIOLI V *et al*
SELLER MJ *et al*. Pseudotrisomy 13 and autosomal recessive holoprosencephaly: *short report*, 970
see also DOLK H and SELLER MJ
SELLINGER B *see* DRISCOLL DA *et al*
SERES-SANTAMARIA A *et al*. Two sibs with cleft palate, ankyloblepharon, alveolar synechiae, and ectodermal defects: a new recessive syndrome? *brief paper*, 793
SERVILLE F *see* LACOMBE D *et al*
SESHIA SS *see* HILDES E *et al*
SEUFERT-SATOMI U *see* TRAUTMANN U *et al*
SEURIN D *see* SCHNEID H *et al*
SHABTAI F *et al*. Chromosome 20 long arm deletion in an elderly malformed man: *short report*, 171
SHAH VC *see* RADHAKRISHNA U *et al*
SHAIKH S *et al*. The dopamine D3 receptor gene: no association with bipolar affective disorder, 308
SHAW DJ *et al*. A study of DNA methylation in myotonic dystrophy, 189
see also MACMILLAN JC *et al*
SHEN Y *see* CALLEN DF *et al*
SHERIDAN E *et al*. Investigation of mutations of the p53 tumour suppressor gene in fresh and archival ovarian cancer material, 335 *abs*
SHICKLE D, HARVEY I. 'Inside-out', back-to-front: a model for clinical population genetic screening, 580
SHIH L-Y *see* ROBINSON WP *et al*
SHILOH Y *see* KLEIMAN S *et al*
SHOKEIR MHK *see* ADAM S *et al*
SHOULSON I *see* MACDONALD ME *et al*
SHRIMPTON AE *et al*. Presymptomatic testing for autosomal dominant spinocerebellar ataxia type 1: *short report*, 616
SHUTT P *see* HERNANDEZ D *et al*
SIGGERS D *see* JOUET M *et al*
SILVER A *et al*. A pilot population carrier screening programme for cystic fibrosis, 343 *abs*
SIMPSON S *see* BARRON LH *et al*
SIMPSON SA *et al*. Huntington's disease in Grampian region: correlation of the CAG repeat number and the age of onset of the disease, 1014
Predictive testing for Huntington's disease: after the gene, 1036
see also REIK W *et al*
SINGH SM *see* RODENHISER DI *et al*
SKIRTON H *see* CONNOR JM *et al*
SKJÖRTEN F *see* ÖRSTAVIK KH *et al*
SKRAASTAD MI *see* DE ROOIJ KE *et al*
SLANEY S *et al*. X linkage in boomerang dysplasia, 341 *abs*
SLATTER R *see* MAHER ER *et al*
SLAUGENHAUPT SA *see* BLUMENFELD A *et al*
SLOMSKI R *see* WILL K *et al*
SMALL KW *see* PERICAK-VANCE MA *et al*
SMEETS DFCM *see* SMITS APT *et al*
SMEETS E *see* FRYNS J-P *et al*
SMITH DI *see* PERICAK-VANCE MA *et al*
SMITH JA *see* YATES JRW *et al*
SMITH RA, GARDNER-MEDWIN D. Orofaciodigital syndrome type III in two sibs: *brief paper*, 870
SMITS APT *et al*. The fragile X syndrome: no evidence for any recent mutations, 94
SMOOKER PM *see* DIANZANI I *et al*
SNELL RG *see* MACMILLAN JC *et al*
SNODGRASS SR *see* MACDONALD ME *et al*
SOEKARMAN D, FRYNS JP. Hypohidrotic ectodermal dysplasia, central nervous system malformation, and distinct facial features: confirmation of a distinct entity? *short report*, 245
SOLANKI JV *see* RADHAKRISHNA U *et al*
SOLTAN H *see* ADAM S *et al*
SOMER M. Diagnostic criteria and genetics of the PEHO syndrome, 932
SONATI MF *see* MARTINS CSB *et al*
SOON GM *see* KNIGHT LA *et al*

SOUSA A *see* DRUGGE U *et al*
SPAANS-VAN DER BIJL T *see* SMITS APT *et al*
SPAZIALE A *see* CAMERA G *et al*
SPEER A *see* LENK U *et al*
SPENCE N *see* KREMER B *et al*
SPENCER K, CARPENTER P. Preliminary results of a prospective study of Down's syndrome screening using Free beta hCG, 336 *abs*
SPERLI D *et al*. Long survival of a patient with Marshall-Smith syndrome without respiratory complications: *brief paper*, 877
SPINNER N *see* CALLEN DF *et al*
SQUITIERI F *see* DI MAIO L *et al*
GOLDBERG YP *et al*
KREMER B *et al*
SRINIDHI J *see* MACDONALD ME *et al*
STACEY M *see* HERNANDEZ D *et al*
MOSS C *et al*
STAJICH JM *see* PERICAK-VANCE MA *et al*
STANCZAK H *see* BOCIAN E *et al*
STANNARD KJD *et al*. Mutation analysis in families with hyperkalaemic periodic paralysis, 348 *abs*
STATHAM H *see* GREEN J *et al*
STEFANINI M *see* LEHMANN AR *et al*
STEFANOVA M *see* VALKOVA G AND STEFANOVA M
STEINMEYER K *see* KOCH MC *et al*
STILLER CA *see* MORRIS JA *et al*
STIRLING JL *see* HARMON DL *et al*
STOFFEL M *et al*. Glucokinase gene mutation in type 2 diabetes mellitus, 333 *abs*
STOLL C *et al*. Trisomy 9 mosaicism in two girls with multiple congenital malformations and mental retardation: *short report*, 433
STRISCIUGLIO P *see* SPERLI D *et al*
STURHMANN M *see* WILL K *et al*
SUCHOWERSKY O *see* ADAM S *et al*
SUFFYS P *see* LLERENA JC *et al*
SUMMERS A *see* ADAM S *et al*
SUMMERS AM *see* FARRELL SA *et al*
SUSLAK L *see* ROBINSON WP *et al*
SUTHERLAND GR, RICHARDS RI. Dynamic mutations on the move: *review article*, 978
see also CALLEN DF *et al*
SUTHERS GK *et al*. Molecular diagnosis of myotonic dystrophy: *letter*, 533
SUZUMORI K *et al*. Prenatal diagnosis of a hypermethylated full fragile X mutation in chorionic villi of a male fetus: *brief paper*, 785
SWEATMAN A *et al*. A new restriction fragment length polymorphism at the DXS101 locus allows carrier detection in a family with X linked agammaglobulinaemia: *short report*, 512
SWENSSON O *see* MEINDL A *et al*
SYKES B *see* WALLIS GA *et al*
SZYMONOWICZ W *see* FARRELL SA *et al*

TAJIK JA *see* MICHELS VV *et al*
TAKAO A *see* BURN J *et al*
TAM P *see* CHUNG E *et al*
TAN M *see* KNIGHT LA *et al*
TAYLOR AMR *see* HERNANDEZ D *et al*
TEEBI AS. Limb/pelvis/uterus-hypoplasia/aplasia syndrome: *letter*, 797
TELENIUS H *see* GOLDBERG YP *et al*
KREMER B *et al*
TESHIMA IE *see* DIXON JW *et al*
THAKKER N *see* EVANS DGR *et al*
THARAPEL AT *see* GORDON PL *et al*
THEILMANN J *see* ADAM S *et al*
GOLDBERG YP *et al*
KREMER B *et al*
THIBODEAU SN *see* MICHELS VV *et al*
THIES U *et al*. Attitudes of neurologists, psychiatrists, and psychotherapists towards predictive testing for Huntington's disease in Germany, 1023
THIRY P *see* FRYNS J-P *et al*
THOMPSON AF *see* LEHMANN AR *et al*
TIETZE HU *see* TRAUTMANN U *et al*
TOMMERUP N. Mendelian cytogenetics. Chromosome rearrangements associated with mendelian disorders: *review article*, 713
TOMMERUP N *et al*. A cytogenetic survey in Menkes disease: implications for the detection of chromosomal rearrangements in X linked disorders: *short report*, 314
Sex dependent transmission of Beckwith-Wiedemann syndrome associated with a reciprocal translocation t(9;11)(p11.2;p15.5): *brief paper*, 958
TØNNESEN T *see* TOMMERUP N *et al*
TORRES MP *see* KOUSSEFF BG *et al*
TORY K *see* RICHARDS FM *et al*
TOWE C *see* COOPER PJ *et al*
TRAUTMANN U *et al*. Simultaneous de novo interstitial deletion of 16q21 and intercalary duplication of 19q in a retarded infant with minor dysmorphic features: *short report*, 330
TREMBATH RC *et al*. A splice site mutation of the COL4A5 gene detected by DGGE causes Alport syndrome, 334 *abs*
TRENT RJ *see* MOTUM PI *et al*
TROFATTER JA *see* BLUMENFELD A *et al*
DI MAIO L *et al*
TSANACLIS AM *see* PASSOS-BUENO MR *et al*
TÜMER Z *see* TOMMERUP N *et al*
TURCO AE *et al*. Molecular genetic diagnosis of autosomal dominant polycystic kidney disease in a newborn with bilateral cystic kidneys detected prenatally and multiple skeletal malformations, 419
TURNER G *see* OGATA T *et al*
RICHARDS FM *et al*
TURPIN JC *see* BENSON MD *et al*
TYLER-SMITH C *see* OGATA T *et al*

UNITED KINGDOM HUNTINGTON'S DISEASE PREDICTION CONSORTIUM *see* SIMPSON SA *et al*
UPTON CJ, YOUNG ID. Severe developmental delay and multiple strawberry naevi: a new syndrome? *short report*, 883
URIOSTE M *see* MARTNEZ-FRIAS M-L *et al*

VAINZOF M *see* PASSOS-BUENO MR *et al*
ZATZ M *et al*
VALDEZ H *see* OROZCO L *et al*
VALERIO D *et al*. A new case of partial trisomy 19q (q13.2→qter) owing to an unusual maternal translocation: *brief paper*, 697
VALERO A *see* PERAL B *et al*
VALKOVA G, STEFANOVA M. Reproductive possibilities for balanced translocation (14) carriers in families with partial trisomy of proximal 14q: *short report*, 73
VALLADA H *see* ZATZ M *et al*
VAN DEN BERGHE H *see* DECRUYENAERE M *et al*
FRYNS J-P *et al*
VAN DER WEIDEN RMF. George Huntington: the man behind the eponym: *letter*, 1042
VAN DIJK M *see* DE DIE-SMULDERS CEM *et al*
VAN DUIJN CM *et al*. Genetic transmission of Alzheimer's disease among families in a Dutch population based study, 640
VAN HAUWAERT J *see* BUNTINCX IM *et al*
VAN HEMEL JO *see* DE VRIES BBA *et al*
VAN MALDERGEM L *see* VERLOES A *et al*
VAN OMMEN G-JB *see* DE ROOIJ KE *et al*
NICHOLSON LVB *et al*
VAN OOST BA *see* SMITS APT *et al*
VAN OVERMEIRE B *see* BUNTINCX IM *et al*
VAN ROY B *see* CALLEN DF *et al*
VAN THIENEN M-N *see* CALLEN DF *et al*
VANAGAITE L *see* KLEIMAN S *et al*
VANCE JM *see* PERICAK-VANCE MA *et al*
VARONE P *see* GIMELLI G *et al*
VAZQUEZ M-P *see* SCHNEID H *et al*
VEGTER-VAN DER VLIS M *see* DE ROOIJ KE *et al*
VEKEMANS M *see* MORICHON-DELVALLEZ N *et al*
VERLOES A. Iris coloboma, ptosis, hypertelorism, and mental retardation: Baraitser-Winter syndrome or Noonan syndrome? *short report*, 425
VERLOES A *et al*. Brachymorphism-onychodysplasia-dysphalangism syndrome: *short report*, 158
VETRIE D. Isolation of the defective gene in X linked agammaglobulinaemia: *annotation*, 452

VIEIRA AHG *see* ZATZ M *et al*
VIETO E *see* WARD J *et al*
VILJOEN D *see* WALLIS GA *et al*
VINKEN L *see* FRYNS J-P *et al*
VINTINER GM *et al*. Exclusion of candidate genes from a role in cleft lip with or without cleft palate: linkage and association studies, 773
VITÉZ M *see* CZEIZEL AE *et al*
VONSATTEL JP *see* MACDONALD ME *et al*

WADEY R *et al*. Isolation of a new marker and conserved sequences close to the DiGeorge syndrome marker HP500 (D22S134), 818
see also BURN J *et al*
WAGSTAFF J *see* ROBINSON WP *et al*
WALD NJ *see* CUCKLE HS *et al*
WALLACE AJ *et al*. Visualisation of microsatellite repeats by silver staining, 346 *abs*
WALLEY AJ *et al*. Gaucher's disease in the United Kingdom: screening non-Jewish patients for the two common mutations, 280
WALLIS GA *et al*. Osteogenesis imperfecta type III: mutations in the type I collagen structural genes, COL1A1 and COL1A2, are not necessarily responsible, 492
WALLIS Y *et al*. Detection of germline mutations in the APC gene in FAP families, 333 *abs*
WALTHER J-U *et al*. Survival in 'lethal' osteogenesis imperfecta: a case report of concordant twins, 337 *abs*
WANG C *et al*. Mild phenotypic manifestation of a 7p15.3p21.2 deletion: *short report*, 610
WANG F, LI Y. A new stable human dicentric chromosome, tdic(4;21)(p16;q22), in a woman with first trimester abortion: *brief paper*, 696
WARBURG M. Classification of microphthalmos and coloboma, 664
WARD J *et al*. Acrocephalopolysyndactyly, pentalogy of Fallot, and hypoacusis in a patient with de novo reciprocal translocation involving the short arm of chromosome 1 and the long arm of chromosome 18: 46,XX,t(1;18)(p31;q11): *short report*, 438
WARNER JP *et al*. Linkage studies in X linked Emery-Dreifuss muscular dystrophy: a new microsatellite marker localises a key recombinant, 345 *abs*
see also BARRON LH *et al*; YATES JRW *et al*
WARREN ST *see* SMITS APT *et al*
WATERS JJ *et al*. Rapid confirmation of trisomy 18 on interphase cells from venous and cardiac blood smears using a directly fluoresceinated 18 centromere specific probe, 336 *abs*
WATKISS E *et al*. X inactivation studies in adrenoleucodystrophy, 342 *abs*
WATKISS RE *et al*. Is skewed X inactivation responsible for symptoms in female carriers for adrenoleucodystrophy? 651
WATSON A *see* JACOBS PA *et al*
WAUTERS J *see* CALLEN DF *et al*
WEBB T *see* CHAN C-TJ *et al*
CLAYTON-SMITH J *et al*
WATKISS E *et al*
WEBER C *see* OUDET C *et al*
WEBER JL *see* MÜLLER U *et al*
WEHNERT M *see* SCHRÖDER W *et al*
WEISSENBACH J *see* PASSOS-BUENO MR *et al*
WELCH JL *see* NICHOLSON LVB *et al*
WELCH JP *see* ADAM S *et al*
WESBY-VAN SWAAY E *see* DE VRIES BBA *et al*
WESTON MD *see* DAHL SP *et al*
WEXLER NS *see* MACDONALD ME *et al*
WEYERTS LK *et al*. Prenatal diagnosis of a giant intracranial teratoma associated with pulmonary hypoplasia: *brief paper*, 880
WHELESS JW *see* NORTHRUP H *et al*
WHISENANT E *see* PERICAK-VANCE MA *et al*
WHITE JM *et al*. Frequency and clinical significance of erythrocyte genetic abnormalities in Omanis, 396
WHITE-VAN MOURIK MCA *et al*. Reproductive behaviour after a second trimester termination of pregnancy, 344 *abs*
WHYTE DA *see* MENNIE ME *et al*
WHYTE P *see* ADAM S *et al*
WICK U *see* KNOBLOCH O *et al*
WICKREMASINGHE A *see* WADEY R *et al*
WIENKER T *see* OWEN MJ *et al*
WIENKER TF *see* WOLFF G *et al*
WIGGINS S *see* ADAM S *et al*
WILCOX DE. The teaching of ethical aspects of medical genetics in undergraduate medicine, 334 *abs*
WILDENAUER DB *see* NÖTHEN MM *et al*
WILKIE AOM *see* COLEMAN R *et al*
WILKINSON ML *see* HODGSON SV *et al*
WILL K *et al*. CFTR transcripts are undetectable in lymphocytes and respiratory epithelial cells of a CF patient homozygous for the nonsense mutation R553X, 833
WILLEMS PJ *see* WILLIAMSON M *et al*
WILLIAMS C, WILLIAMSON R. Reliability of single cell PCR, 332 *abs*
WILLIAMS C *et al*. A report on CF carrier frequency among men with infertility owing to congenital absence of the vas deferens: *letter*, 973
WILLIAMS DM *see* CLARKSON PA *et al*
WILLIAMS J *see* OWEN MJ *et al*
WILLIAMSON M *et al*. A 5′ splice site mutation in fucosidosis, 218
WILLIAMSON R *see* WILLIAMS C *et al*
WILLSHAW HA *see* GLASS IA *et al*
WILROY RS *see* GORDON PL *et al*
WILSON D *see* BURN J *et al*
WADEY R *et al*
WILSON DI *et al*. DiGeorge syndrome: part of CATCH 22: *syndrome of the month*, 852
WILSON GN. Aphasia, deafness, or mental retardation: *letter*, 262
WILSON J *see* BARAITSER M *et al*
REARDON W *et al*
WINCHESTER B *see* WILLIAMSON M *et al*
WINCHESTER BG *see* DAVIES JP *et al*
WINTER RM. Molecular diagnosis of myotonic dystrophy: *letter*, 533
Skeletal malformations and polycystic kidney disease: *letter*, 973
see also HOLDER SE *et al*; HOLDER SE and WINTER RM; VINTINER GM *et al*
WITT M *et al*. An improved, non-isotopic method of screening cells from patients with abnormalities of sexual differentiation for Y chromosomal DNA content, 304
WOLF F *see* KOCH MC *et al*
WOLFF G *et al*. On the genetics of mandibular prognathism: analysis of large European noble families, 112
WOO SLC *see* KLEIMAN S *et al*
WOODS CG *see* HERNANDEZ D *et al*
WORLD FEDERATION OF NEUROLOGY RESEARCH GROUP ON HUNTINGTON'S DISEASE. Presymptomatic testing for Huntington's disease: a world wide survey, 1020
WROGEMANN K *see* HILDES E *et al*

YAMAOKA LH *see* PERICAK-VANCE MA *et al*
YAMAUCHI M *see* SUZUMORI K *et al*
YATES J *see* JOUET M *et al*
YATES JRW *et al*. Emery-Dreifuss muscular dystrophy: linkage to markers in distal Xq28, 108
see also RICHARDS FM *et al*; Zhang Y *et al*
YILMAZ E *see* ÖZGÜÇ M *et al*
YOUINGS S *see* JACOBS PA *et al*
YOUNG A *see* MACDONALD ME *et al*
YOUNG E *see* WILLIAMSON M *et al*
YOUNG ID *et al*. A lethal skeletal dysplasia with generalised sclerosis and advanced skeletal maturation: Blomstrand chondrodysplasia? *short report*, 155
see also BUNDEY S and YOUNG ID; JENKINS D *et al*; NEWBURY-ECOB RA *et al*; UPTON CJ AND YOUNG ID

ZACKAI E *see* CALLEN DF *et al*

ZACKAI EH *see* DRISCOLL DA *et al*
GOLDMUNTZ E *et al*
ZAREMBA J *see* YATES JRW *et al*
ZATZ M *et al.* Cosegregation of schizophrenia with Becker muscular dystrophy: susceptibility locus for schizophrenia at Xp21 or an effect of the dystrophin gene in the brain? 131
see also EGGERS S *et al*; PASSOS-BUENO MR *et al*
ZBAR B *see* RICHARDS FM *et al*
ZEIGER K *see* LANDELS EC *et al*
ZELDENRUST S *see* BENSON MD *et al*
ZELDENRUST SR *et al.* RFLP analysis for APP 717 mutations associated with Alzheimer's disease, 476
ZENG F-Y *see* ZENG Y-T *et al*
ZENG Y-T *et al.* A new de novo mutation (A113T) in HMG box of the SRY gene leads to XY gonadal dysgenesis, 655
ZERRES K *et al.* Childhood onset autosomal dominant polycystic kidney disease in sibs: clinical picture and recurrence risk, 583
ZHANG M-L *see* ZENG Y-T *et al*
ZHANG Y *et al.* Genetic mapping of the Kallmann syndrome and X linked ocular albinism gene loci, 923
ZHENG Y *et al.* Prenatal diagnosis from maternal blood: simultaneous immunophenotyping and FISH of fetal nucleated erythrocytes isolated by negative magnetic cell sorting, 1051
ZOLL B *see* KNOBLOCH O *et al*
KOCH MC *et al*
ZOLLINO M *see* KOZÁK L *et al*
ZSCHIESCHE M *see* SCHRÖDER W *et al*
ZUCCOLLO JM *see* NEWBURY-ECOB RA *et al*
YOUNG ID *et al*
ZUFFARDI O *see* BRISCIOLI V *et al*
ROSSI E *et al*
ZWIERLEIN E *see* DE DINECHIN O *et al*

VOLUME 30 · SUBJECT INDEX

Acrocephalopolysyndactyly, pentalogy of Fallot, and hypoacusis, with 46,XX,t(1;18)(p31;q11): *short report*, 438
Adrenoleucodystrophy
  is skewed X inactivation responsible for symptoms in female carriers?, 651
  X inactivation studies, 342 *abs*
Agammaglobulinaemia, X linked
  isolation of defective gene: *annotation*, 452
  new RFLP at DXS101 locus allows carrier detection: *short report*, 512
Airway abnormalities in Jarcho-Levin syndrome, two cases: *brief report*, 875
Al-Awadi/Raas-Rothschild syndrome
  Bedouin patient: *short report*, 62: *letter*, 797
  two Italian sibs and further confirmation of autosomal recessive inheritance: *short report*, 65: *letter*, 797
Åland Island eye disease, genetic mapping to proximal short arm of human X chromosome, 1044
Albright's hereditary osteodystrophy, imprinting, 101
Alport syndrome, splice site mutation of COL4A5 gene detected by DGGE causing, 334 *abs*
Alveolar synechiae, ectodermal defects, ankyloblepharon, and cleft palate, in two sibs, a new recessive syndrome? *brief paper*, 793
Alzheimer's disease
  APP 717 mutations associated with, RFLP analysis, 476
  genetic transmission, Dutch families, 640
Amelogenesis imperfecta, X linked, nonsense mutation in gene, 345 *abs*
Amniocyte and CVS DNA, direct amplification, in rapid diagnosis of infantile spinal muscular atrophy: *short report*, 162
Amyloidotic polyneuropathy, familial
  and transthyretin variant (alanine 49), French family, 117
  and transthyretin variant (alanine 71), French family, 120
Amyotrophic lateral sclerosis, familial, and chromosome 21 loci, absence of linkage between: *short report*, 318
Anaemias, hereditary, Portugal, epidemiology, public health significance, and control, 235
Anderson-Fabry disease, and alpha-galactosidase A, sequence variations, 658
Androgen receptor gene, mutations identified in perineal hypospadias, 198
Aneuploidy, and fragile X syndrome, two cases, 338 *abs*
Angelman syndrome
  developmental delay and ataxia, duplication of chromosome 15: *short report*, 529
  molecular genetic studies, 348 *abs*
  molecular mechanisms, 93 patients, 895
  or Prader-Willi syndrome, uniparental disomy, in patients with an additional small inv dup(15) chromosome, 756
Ankyloblepharon, alveolar synechiae, ectodermal defects, and cleft palate, in two sibs, a new recessive syndrome? *brief paper*, 793
Ankyloblepharon filiforme adnatum, complex chromosome rearrangement: *short report*, 167
Anophthalmia and pituitary abnormalities, and deletion 14(q22.1→q22.3): *short report*, 251
APC gene
  detection of germline mutations, in FAP families, 333 *abs*
  in FAP families, and sporadic colorectal cancer, detection of mutations, 339 *abs*
  germline rearrangement detected by PFGE, 340 *abs*
Aphasia, deafness, or mental retardation: *letter*, 262
APP 717 mutations associated with Alzheimer's disease, RFLP analysis, 476
Arthrogryposis, ophthalmoplegia, and retinopathy, confirmation of new type of arthrogryposis: *short report*, 78
Association versus linkage studies in psychosis genetics: *annotation*, 634; *comment*, 638
Ataxia, familial, deafness, and mental retardation, new form: *brief paper*, 694
Ataxia telangiectasia gene and chromosome 11q22-23, family showing no evidence of linkage between, 135
Attitudes of neurologists, psychiatrists, and psychotherapists towards predictive testing for Huntington's disease, Germany, 1023
Autosomal dominant polycystic kidney disease
  childhood onset, in sibs, clinical picture and recurrence risk, 583
  diagnosis by genetic markers and ultrasonography, 332 *abs*
  estimating locus heterogeneity, Spanish population, 910
  phenotype and genotype heterogeneity, 333 *abs*
Autosomal dominant spinocerebellar ataxia type 1, presymptomatic testing: *short report*, 616
Autosomal recessive disorders
  complex consanguinity, autozygosity mapping: *letter*, 798
  and complex consanguinity, homozygosity mapping, 345 *abs*
  low segregation ratios: *annotation*, 449
Autozygosity mapping, complex consanguinity, and autosomal recessive disorders: *letter*, 798

Balding, premature, profound mental retardation, characteristic facies, muscular hypotrophy and small patellae, two unrelated male patients: *short report*, 319
Baraitser-Winter syndrome, or Noonan syndrome: iris coloboma, ptosis, hypertelorism, and mental retardation: *short report*, 425
Beale's syndrome, link with 5q deletion in FAP, 340 *abs*
Becker muscular dystrophy
  carrier detection by in situ hybridisation, 347 *abs*
  investigation based on accurate carrier detection, 347 *abs*
  non-isotopic analysis of SSCP in exon 13 region of human dystrophin gene, 951
  prevalence and incidence, Slovenia, 344 *abs*
  and schizophrenia, 131
Becker type generalised myotonia, autosomal recessive, evidence for genetic homogeneity, 914
Beckwith-Wiedemann syndrome
  associated with reciprocal translocation t(9;11)(p11.2;p15.5), sex dependent transmission: *brief paper*, 958
  and parental allele specific methylation of IGF-II gene, 353
Bedouin infant, limb/pelvis-hypoplasia/aplasia syndrome: *short report*, 62: *letter*, 797
Benign hereditary chorea, identification of CAG repeat in Huntington's disease gene (IT15), N Ireland, 1012
Beta-hexosaminidase alpha subunit gene, exon 1, in two mutations in late infantile Tay-Sachs disease, 123
Bipolar affective disorder, no association with dopamine D3 receptor gene, 308

Blaschko's lines, and epidermal mosaicism, 752
Blepharophimosis, heart defect, hypothyroidism, mental retardation syndrome (Young-Simpson syndrome), parental consanguinity: *short report*, 255
Blepharophimosis, ptosis, epicanthus inversus syndrome, example favouring location at 3q2: *letter*, 86
Bloch-Sulzberger syndrome (incontinentia pigmenti): *syndrome of the month*, 53
Blomstrand chondrodysplasia, presenting with lethal skeletal dysplasia with generalised sclerosis and advanced skeletal maturation? *short report*, 155
Boomerang dysplasia, X linkage, 341 *abs*
Brachymorphism-onychodysplasia-dysphalangism syndrome: *short report*, 158
British Medical Genetics Conference, Nottingham, 23-25 September 1992, abstracts, 332
Bronchiectasis and immotile nasal cilia, associated with Usher syndrome type I, two brothers: *short report*, 253
Bullous aplasia cutis congenita, and epidermal naevi, in a neonate: *brief paper*, 962

CAG repeat in Huntington's disease, 1003
  and age of onset, Scotland, 1014
  gene (IT15), in benign hereditary chorea, 1012
CAG repeat length, in Huntington's disease chromosomes, 982
Campomelic dysplasia, evidence of autosomal dominant inheritance: *brief paper*, 683
Caroli's disease, link with 5q deletion in FAP, 340 *abs*
Carpotarsal osteochondromatosis, dominant: *brief paper*, 704
CATCH 22
  chromosome 22q11 microdeletions: *editorial*, 801
  DiGeorge syndrome: *syndrome of the month*, 852
Central nervous system malformation, hypohidrotic ectodermal dysplasia, and distinct facial features: confirmation of a distinct entity? *short report*, 245
Centromere involvement, elucidation in inversion (13) by fluorescent in situ hybridisation: *short report*, 414
Cerebellar ataxia and ectodermal dysplasia in brothers: *short report*, 515
Cerebral palsy, parental age and genetic mutation, 44
Charcot-Marie-Tooth disease, X linked, linkage study using highly informative loci, 345 *abs*
Chemical cleavage of mismatch to screen exons of phenylalanine hydroxylase gene in PKU, 228
Chiasma frequency, quadrivalent orientation, and first meiotic segregation in a human male reciprocal translocation heterozygote t(15;20)(q13;q13.1), 336 *abs*
Choanal atresia, bilateral, cleft palate, curly hair, and congenital hypothyroidism, syndromic association: *short report*, 427
Chorea, benign hereditary, identification of CAG repeat in Huntington's disease gene (IT15), N Ireland, 1012
Chorionic villus DNA, direct amplification, in rapid diagnosis of infantile spinal muscular atrophy: *short report*, 162
Chromosome abnormalities
  2(p11-p13) deletion: *short report*, 604
  2q, terminal deletions, is there a consistent phenotype? 338 *abs*
  (4;21)(p16;q22), tdic, with first trimester abortion: *brief paper*, 696
  5q deletion
    in familial adenomatous polyposis, possible link with Caroli's disease, 369
    in FAP, and Caroli's disease and Beale's syndrome, 340 *abs*
  7;15 balanced translocation, congenital nystagmus cosegregating: *short report*, 526
  7p15.3p21.2 deletion, mild phenotypic manifestation: *short report*, 610
  7q36→7qter terminal deletion, and holoprosencephaly and sacral agenesis: *short report*, 521
  9
    extra G positive band on long arm: *short report*, 613
    tuberous sclerosis locus, 224
  9p deletion, and sex reversal: *short report*, 518
  9q11-q33, markers, and linkage analysis of pyloric stenosis, 393
  10q23, interstitial deletion, new case and review: *short report*, 248
  10qter trisomy confirmed by in situ hybridisation: *short report*, 601
  11q22-23, and ataxia telangiectasia gene, family showing no evidence of linkage between, 135
  13, inversion, elucidation of centromere involvement by fluorescent in situ hybridisation: *short report*, 414
  15, inv dup, additional, in Angelman or Prader-Willi syndrome, explained by uniparental disomy, 756
  15q11-13 duplication, similarities to Angelman syndrome: *short report*, 529
  16 deletion
    duplication 19q, in 46,XX,del(16)(q13.08-21.05),dup(19)(q13.13-13.2) karyotype, dysmorphic features and retardation: *short report*, 330
    interstitial long arm, high resolution mapping, relationship to phenotype, 828
  17q, probable de novo duplication, in Klinefelter's syndrome: *short report*, 436
  18
    ring, transmission from mother with 46,XX/47,XX,+r(18) mosaicism to daughter, resulting in 46,XX,r(18) karyotype: *brief paper*, 964
    triple structural mosaicism, in child with MR/MCA syndrome and abnormal skin pigmentation: *short report*, 614
  19q (q13.2→qter), partial trisomy, unusual maternal translocation: *brief paper*, 697
  20 long arm deletion, in elderly malformed man: *short report*, 171
  21, loci, and familial amyotrophic lateral sclerosis, absence of linkage between: *short report*, 318
  22q11 microdeletions: *editorial*, 801
  22q11 microdeletions
    and conotruncal anomaly face syndrome, 822
    in DiGeorge and velocardiofacial syndromes, prevalence, implications for genetic counselling and prenatal diagnosis, 813
    in patients with congenital conotruncal cardiac defects, 807
  46,XX/69,XX diploid-triploid mixoploidy, with hypothyroidism and precocious puberty: *brief paper*, 966
  46,XX/69,XXX mixoploidy, resulting from second polar body incorporation into blastomere: *short report*, 597
  46,XX,t(1;18)(p31;q11), with acrocephalopolysyndactyly, pentalogy of Fallot, and hypoacusis: *short report*, 438
  biochemical screening, 339 *abs*
  de novo complex rearrangement in ankyloblepharon filiforme adnatum: *short report*, 167
  location of mutations causing disease: *review article*, 1
  and neural tube defects, biochemical screening, *abs* 336
  non-disjunction, centromere map analysis, 335 *abs*
  rearrangements associated with mendelian disorders: *review article*, 713
  rearrangements leading to interstitial telomeric sequences, types, stability, and phenotypic consequences, 926
  t(13;18)(13p18p;13q18q), resulting in three different adverse pregnancy outcomes: *short report*, 417
  and Williams-Beuren syndrome: *letter*, 886
  X
    inactivation, and X linked disease diagnosis, females, 177
    new mental retardation syndrome mapping to pericentromeric region, 337 *abs*
    proximal short arm, in Åland Island eye disease, 1044

Xp21.1–Xp22.3, evidence for linkage to non-specific X-linked mental retardation: *brief paper*, 866
Xp21 susceptibility locus for schizophrenia, 131
Xp22.3 terminal deletion, analysis in patient with six monogenic disorders, implications for mapping of X linked ocular albinism, 838
Xq28, distal, linkage markers, and Emery-Dreifuss muscular dystrophy, 108
XY gonadal dysgenesis, new de novo mutation (A113T) of SRY gene leads to, 655
Y, DNA content, abnormalities of sexual differentiation, improved method of screening cells, 304
Yp localisation of gene(s) for Turner stigmata, 918
Cleft lip with or without cleft palate, exclusion of candidate genes, linkage and association studies, 773
Cleft palate
ankyloblepharon, alveolar synechiae, and ectodermal defects, in two sibs, a new recessive syndrome? *brief paper*, 793
bilateral choanal atresia, curly hair, and congenital hypothyroidism, syndromic association: *short report*, 427
Clefting, ear anomalies, and limb reduction defects, new autosomal recessive condition? 342 *abs*
Clinical genetics, specialty, European Society of Human Genetics survey, 147
Coarctation of the aorta, interrupted aortic arch, and hypoplastic left heart syndrome, in three generations: *short report*, 328
Cockayne's syndrome, correlation of clinical features with cellular sensitivity of RNA synthesis to UV radiation, 679
COL2A1, in osteoarthritis, molecular analysis, 348 *abs*
COL3A1 IVS7 exon skipping mutation, resulting in Ehlers-Danlos syndrome type IV, 376
COL4A5 gene, splice site mutation, detected by DGGE, causes Alport syndrome, 334 *abs*
Collagen, type II, alpha-1(II) chain, substitution of glycine 997 by serine in, and spondyloepiphyseal dysplasia congenita, 27
Coloboma, classification, 664
Colorectal cancer, sporadic, FAP families, APC gene mutations, 339 *abs*
Cone and rod dysfunction (Åland Island eye disease), genetic mapping to proximal short arm of human X chromosome, 1044
Cone-rod dystrophy, associated with subretinal neovascularisation and gingival dystrophy, 342 *abs*
Congenital heart malformation in Yunis-Varón syndrome: *brief paper*, 788
Congenital malformation, Emilia Romagna birth defects register, 344 *abs*
Conotruncal anomaly face syndrome is associated with chromosome 22q11 microdeletions, 822
Conotruncal cardiac defects, congenital, and 22q11 microdeletions, 807
Consanguinity, complex, and autosomal recessive disorders
autozygosity mapping: *letter*, 798
homozygosity mapping, 345 *abs*
Correction, 88, 528
Costello syndrome, and cutis laxa: *correspondence*, 622
Counselling pitfalls, Romano-Ward syndrome, consanguinity, cardiac arrest, hearing impairment, and ECG abnormalities: *short report*, 325: *letter*, 886
Craniosynostosis, and absent fibula, 25 year follow up: *letter*, 445
Crigler Najjar syndrome type I, DNA markers for analysis, 346 *abs*
Curly hair, cleft palate, bilateral choanal atresia, and congenital hypothyroidism, syndromic association: *short report*, 427
Cutis laxa and the Costello syndrome: *letter*, 622
Cystic fibrosis
absence, Asia, occurrence of homozygous S549N mutation in inbred Pakistani family: *short report*, 164
antenatal carrier screening, 343 *abs*
carrier frequency among men with infertility owing to congenital absence of the vas deferens: *letter*, 973
carrier testing availability, young people's reactions, 538
CFTR transcripts are undetectable in lymphocytes and respiratory epithelial cells homozygous for nonsense mutation R553X, 833
and deafness: *letter*, 621
diagnosis, preimplantation, by DNA amplification from single cells, 332 *abs*
and fetal echogenic bowel, in low risk fetuses, 333 *abs*
frequency of ΔF508 in Mexican sample of patients, 501
in Grampian, 343 *abs*
mild pulmonary but severe hepatic disease, and CFTR frameshift mutation: *letter*, 446
mutations, prevalence in Grampian region of Scotland: *short report*, 316
pilot population carrier screening programme, 343 *abs*
prenatal carrier screening
new approach: *letter*, 86; *letter*, 621
two approaches: *letter*, 621
prenatal screening, psychological effects on carriers and their partners, 543
relative risk and risky relatives, 344 *abs*
severe, in child homozygous for G542 nonsense mutation in CFTR gene: *letter*, 621
severity of chest disease in relation to genotypes: *letter*, 709
unusual family, 343 *abs*
when homozygous for mild mutation R117H, male infertility as only presenting sign: *letter*, 797
Cytogenetics editor, Professor Pat Jacobs standing down, Professor Albert Schinzel taking over, 1043

Deafness
aphasia, or mental retardation: *letter*, 262
and cystic fibrosis: *letter*, 621
familial ataxia, and mental retardation, new form: *brief paper*, 694
Delayed speech development, facial asymmetry, strabismus, and transverse ear lobe creases, new syndrome: *short report*, 76
Deletion
of chromosome 2(p11-p13): *short report*, 604
de novo interstitial, of 16q21 and duplication of 19q, with dysmorphic features in retarded infant: *short report*, 330
interstitial, of chromosome 10q23, new case and review: *short report*, 248
terminal, 7q36→7qter, and holoprosencephaly and sacral agenesis: *short report*, 521
5q, in FAP, possible link with Caroli's disease, 369
7p15.3p21.2, mild phenotypic manifestation: *short report*, 610
9p, and sex reversal: *short report*, 518
14(q22.1→q22.3), associated with anophthalmia and pituitary abnormalities: *short report*, 251
Desmoid tumours, and FAP, with congenital skull defect, 342 *abs*
Developmental delay, severe, and multiple strawberry naevi, a new syndrome? *short report*, 883
Diabetes mellitus, type I insulin dependent, contribution of genetic factors to pathogenesis: *letter*, 533
Diabetes mellitus type 2, glucokinase gene mutation, 333 *abs*
Diastrophic dysplasia, prenatal diagnosis with polymorphic DNA markers, 265
Dicentric chromosome, new stable human, tdic(4;21)(p16;q22), in woman with first trimester abortion: *brief paper*, 696
DiGeorge syndrome
and 22q11 microdeletions: *editorial*, 801, 807
prevalence, implications for genetic counselling and prenatal diagnosis, 813
historical review of clinical and cytogenetic features: *review article*, 803

marker HP500 (D22S134), isolation of new marker and conserved sequences close to, 818
part of CATCH 22: *syndrome of the month*, 852
Dihydropteridine reductase gene, in tetrahydrobiopterin deficiency, two new mutations, 465
Dilated cardiomyopathy, idiopathic, dystrophin analysis, 955
Dinucleotide repeat polymorphisms and von-Hippel-Lindau disease on chromosome 3p25-26, genetic mapping, 487
Diploid-triploid mixoploidy (46,XX/69,XX), with hypothyroidism and precocious puberty: *brief paper*, 966
Disease gene mapping, in isolated human populations, Finland, 857
DNA
markers, polymorphic, in prenatal diagnosis of diastrophic dysplasia, 265
methylation
at *D4S95*, and age at onset in Huntington's disease, 185
in myotonic dystrophy, study, 189
six polymorphisms in LDL receptor gene, 273
technique for Hunter syndrome (MPS II) carrier detection, 210
Dominant carpotarsal osteochondromatosis: *brief paper*, 704
Dopamine D3 receptor gene
association between schizophrenia and homozygosity: *letter*, 708
no association with bipolar affective disorder, 308
Down's syndrome
maternal serum screening, East Anglia, 338 *abs*
screening using Free beta hCG, 336 *abs*
Duchenne muscular dystrophy
alternative splicing of dystrophin mRNA complicates carrier determination, 206
carrier detection by in situ hybridisation, 347 *abs*
carrier risks, women's understanding, 562
different molecular mutations in male sibs, 347 *abs*
discrepancy in estimation of relative male and female mutation rates, 333 *abs*
impact of genetic counselling after neonatal screening for, 670
investigation based on accurate carrier detection, 347 *abs*
non-isotopic analysis of SSCP in exon 13 region of human dystrophin gene, 951
preimplantation diagnosis, development and validation of laboratory procedures, 903
prevalence and incidence, Slovenia, 344 *abs*
Duplication 12q (12q13–q24.2), mosaicism, dysmorphic male infant: *short report*, 70
Duplication of 19q, and interstitial deletion of 16q21, with dysmorphic features and retardation: *short report*, 330
Dwarfism, short limbed, genital hypoplasia, sparse hair, and vertebral anomalies: Ellis-van Creveld syndrome variant? *short report*, 322
DXS207 locus, and retinoschisis disease gene, 300
Dysautonomia, familial, exclusion from more than 60% of genome, 47
Dyskeratosis congenita, three additional families show linkage to locus in Xq28: *short report*, 618
Dystrophin analysis in idiopathic dilated cardiomyopathy, 955
Dystrophin gene
deletions, rapid analysis shows variable electrophoretic mobility, 60
effect on brain in schizophrenia, 131
SSCP in exon 13 region, non-isotopic analysis, 951
Dystrophin mRNA, alternative splicing complicates carrier determination, report of DMD family, 206

Ear anomalies, clefting, and limb reduction defects, new autosomal recessive condition? 342 *abs*
Ectodermal defects, ankyloblepharon, alveolar synechiae, and cleft palate, in two sibs, a new recessive syndrome? *brief paper*, 793
Ectodermal dysplasia and cerebellar ataxia in brothers: *short report*, 515
Ehlers-Danlos syndrome type IV
is caused by substitution of glycine 661 by arginine in type III collagen: *brief paper*, 690
T^{+6} to C^{+6} mutation of COL3A1 IVS7 causes exon skipping resulting in, 376
Ellis-van Creveld syndrome, variant, short limbed dwarfism, genital hypoplasia, sparse hair, and vertebral anomalies: *short report*, 322
Emery-Dreifuss muscular dystrophy
linkage to markers in distal Xq28, 108
X linked, linkage studies, 345 *abs*
Emilia Romagna birth defects register, goals and accomplishments, 344 *abs*
Epidermal mosaicism and Blaschko's lines, 752
Epidermal naevi, and bullous aplasia cutis congenita, in a neonate: *brief paper*, 962
Erythrocyte genetic abnormalities, frequency and clinical significance, Omanis, 396
Ethical aspects of medical genetics, teaching in undergraduate medicine, 334 *abs*
Ethical and social issues in presymptomatic testing for Huntington's disease, European Community collaborative study, 1028
Ethics of human genome analysis: *conference report*, 257
European Society of Human Genetics survey, the specialty of clinical genetics, 147

ΔF508, frequency in Mexican sample of cystic fibrosis patients, 501
Facial asymmetry, delayed speech development, strabismus, and transverse ear lobe creases, new syndrome: *short report*, 76
Facies, characteristic, profound mental retardation, premature balding, muscular hypotrophy and small patellae, two unrelated male patients: *short report*, 319
Facioscapulohumeral muscular dystrophy, aspects of genetic counselling, acceptance of preclinical diagnosis, and fitness, 589
Familial adenomatous polyposis
5q deletions, possible link with Caroli's disease, 369
detection of germline mutations in APC gene, 333 *abs*
preclinical diagnosis, Wales, 340 *abs*
and sporadic colorectal cancer, APC gene mutations, 339 *abs*
variant, with congenital skull defect and recurrent abdominal desmoids, 342 *abs*
Familial adenomatous polyposis coli
5q deletions, link with Caroli's disease and Beale's syndrome, 340 *abs*
evaluation of molecular genetic diagnosis, 675
Familial amyloidotic polyneuropathy, Sweden, pedigree analysis, 388
Familial dysautonomia, exclusion from more than 60% of genome, 47
Familial hypercholesterolaemia, six DNA polymorphisms in LDL receptor gene, 273
Familial ovarian cancer register, psychosocial issues raised, 575
Fanconi anaemia, dilemma of two cell populations, 340 *abs*
Fibrillin gene FBN1, screening for causal mutations in Marfan syndrome, 348 *abs*
Fibrodysplasia ossificans progressiva, in three generation family: *brief paper*, 687
Fibula, absent, and craniosynostosis, 25 year follow up: *letter*, 445
Filipino beta thalassaemia, high Hb A_2 β° thalassaemia resulting from large deletion of 5′ globin gene region, 240
Forensic medicine and the polymerase chain reaction technique: *review article*, 625
Fragile X mutation
detection, north-east of Scotland, 346 *abs*
families, analysis, 346 *abs*

full, hypermethylated, in chorionic villi of male fetus, prenatal diagnosis: *brief paper*, 785
mental retardation, direct versus indirect molecular diagnosis, 40 German families at risk, 193
population studies, molecular approach, 454
with Prader-Willi-like phenotype, clinical and molecular studies, 761
Fragile X syndrome
and aneuploidy, two cases, 338 *abs*
families in west of Scotland, 341 *abs*
found in Martin Bell family, 341 *abs*
FRAXE site, and X linked mental retardation, 97
guidelines for diagnosis, 410
mutation, origins, 647
no evidence for any recent mutations, 94
specific mutation: *review article*, 978
FRAXE, specific mutation: *review article*, 978
FRAXE site, fragile X syndrome, and X linked mental retardation, 97
Free beta hCG, in Down's syndrome screening, 336 *abs*
Fucosidosis, 5′ splice site mutation, 218

alpha-Galactosidase A, sequence variations in first exon, 658
Gaucher's disease
glucocerebrosidase locus, molecular analysis of lysosomal enzyme: *review article*, 889
UK, screening non-Jewish patients for two common mutations, 280
Gene, disease, mapping, in isolated human populations, Finland: *medical genetics around the world*, 857
Genetic counselling, after neonatal screening for Duchenne muscular dystrophy, impact, 670
Genetic diseases, common, molecular diagnosis, Russia and former USSR: *medical genetics around the world*, 141
Genetic screening, clinical population model, 'inside-out', back-to-front, 580
Genetics
medical
ethical aspects, teaching in undergraduate medicine, 334 *abs*
in reformed NHS and Europe, 334 *abs*
psychosis, association versus linkage studies: *annotation*, 634; *comment*, 638
Genital abnormalities, in males, and mutational screening of Wilms's tumour gene WT1, 767
Genital hypoplasia, short limbed dwarfism, sparse hair, and vertebral anomalies: Ellis-van Creveld syndrome variant? *short report*, 322
Genome
analysis, ethics: *conference report*, 257
human, morbid anatomy, chromosomal location of mutations causing disease: *review article*, 1
Germany, attitudes of neurologists, psychiatrists, and psychotherapists towards predictive testing for Huntington's disease, 1023
Glucocerebrosidase locus, in Gaucher's disease, molecular analysis of lysosomal enzyme: *review article*, 889
Glucokinase gene mutation in type 2 diabetes mellitus, 333 *abs*
Glycine 661, substitution by arginine in type III collagen causes Ehlers-Danlos syndrome type IV: *brief paper*, 690
Glycine 997, substitution by serine in alpha-1(II) chain of type II collagen, and spondyloepiphyseal dysplasia congenita, 27
Gonadal dysgenesis, in XY female, new de novo mutation (A113T) of SRY gene, 655
Gorlin syndrome
and congenital anomalies in medulloblastoma, 337 *abs*
flanking markers for gene for, 345 *abs*
Grampian region of Scotland, prevalence of cystic fibrosis mutations: *short report*, 316

Haemophilia, resulting from interaction of incontinentia pigmenti and factor VIII mutations in female with biased X inactivation, 497
Hajdu-Cheney syndrome, and hydrocephalus: *letter*, 175
HEXA gene intron 9 donor splice site mutation frequently found in non-Jewish Tay-Sachs disease patients from the British Isles, 479
Holland, dynamic mutation in Huntington's disease, 996
Holoprosencephaly
autosomal recessive, and pseudotrisomy 13: *short report*, 970
family showing dominant inheritance and variable expression, 36
and sacral agenesis, in fetus with terminal deletion 7q36→7qter: *short report*, 521
Homozygosity, and schizophrenia, association between, at dopamine D3 receptor gene: *letter*, 708
HP500 marker, DiGeorge syndrome, isolation of new marker and conserved sequences close to, 818
Humeroradioulnar synostosis, in patient with lambdoid synostosis: *short report*, 81
Hunter syndrome (MPS II), carrier detection by biochemical and DNA techniques in families at risk, 210
Huntington, George, the man behind the eponym: *portraits in medical genetics*, 406: *letter*, 1042
Huntington's disease
15th International World Federation of Neurology Workshop, 31 August-3 September 1993, Boston, Massachusetts, USA: *conference report*, 1039
age at onset, and methylation at *D4S95*, 185
associated trinucleotide repeat, Scotland, 1003
CAG repeat length, gametic but not somatic instability, 982
dynamic mutation, increased paternal repeat instability extending to within the normal size range, Dutch patients, 996
gene (IT15), identification of CAG repeat in benign hereditary chorea, N Ireland, 1012
Grampian region, correlation of CAG repeat number and age of onset, 1014
late onset, molecular analysis, 991
lessons learnt from testing for, DNA storage and duplicate sampling: *letter*, 1042
mutation size and age at onset, 1008
N Ireland population, heterozygote frequency, mutation rate, fertility, and genetic fitness, 344 *abs*
ocular motor abnormalities, 344 *abs*
onset symptoms, 510 patients, 289
predictive testing
after the gene, 1036
attitudes of neurologists, psychiatrists, and psychotherapists, Germany, 1023
multiallelic probe pKP1.65, 347 *abs*
in one family, 347 *abs*
perception by young women, preferring uncertainty to certainty?, 557
prenatal testing, demand, attitudes, and psychological assessment, 549
presymptomatic testing
ethical and social issues, European Community collaborative study, 1028
world wide survey, 1020
recurrent mutations causing, familial predisposition, genetic risk to sibs of sporadic cases, 987
significant linkage disequilibrium between locus and markers at loci D4S10, D4S95, and D4S111, Northern Ireland, 1018
specific mutation: *editorial*, 975: *review article*, 978
suicide risk, 293
Huriez, scleroatrophic syndrome of, 342 *abs*
Hydrocephalus
in Hajdu-Cheney syndrome: *letter*, 175
X linked, refining genetic location of gene within Xq28, 214
Hyper IgM syndrome (HIGM1), X linked form, mapping, 202

Hyperkalaemic periodic paralysis, mutation analysis in families, 348 *abs*
Hypermethylated full fragile X mutation in chorionic villi of a male fetus, prenatal diagnosis: *brief paper*, 785
Hypertelorism, iris coloboma, ptosis, and mental retardation: Baraitser-Winter syndrome or Noonan syndrome? *short report*, 425
Hypoacusis, acrocephalopolysyndactyly, and pentalogy of Fallot, with 46,XX,t(1;18)(p31;q11): *short report*, 438
Hypohidrotic ectodermal dysplasia, central nervous system malformation, and distinct facial features: confirmation of a distinct entity? *short report*, 245
Hypomelanosis of Ito associated with mosaicism for trisomy 7 and apparent 'pseudomosaicism' at amniocentesis: *brief paper*, 783
Hypoplastic left heart syndrome, coarctation of the aorta, and interrupted aortic arch, in three generations: *short report*, 328
Hypospadias, perineal, androgen receptor gene mutations identified, 198
Hypothyroidism, congenital, cleft palate, bilateral choanal atresia, and curly hair, syndromic association: *short report*, 427
Hypotrophy, muscular, profound mental retardation, characteristic facies, premature balding, and small patellae, two unrelated male patients: *short report*, 319

ICF syndrome with variable expression, in sibs: *short report*, 429
Immunodeficiency, centromeric instability, facial anomalies, with variable expression, in sibs: *short report*, 429
Inbreeding, effect, prospective study, 1986-1992, 334 *abs*
Incontinentia pigmenti
clinical and genetic study, 341 *abs*
interaction with factor VIII mutations in female with biased X inactivation, resulting in haemophilia, 497
Incontinentia pigmenti (Bloch-Sulzberger syndrome): *syndrome of the month*, 53
Ingterstitial telomeric sequences, types, stability, and phenotypic consequences of chromosome rearrangements leading to, 926
Insulin-like growth factor II gene, methylation, and Beckwith-Wiedemann syndrome, 353
Insulin resistance, resembling leprechaunism, 5 sibs, 470
Interrupted aortic arch, coarctation of the aorta, and hypoplastic left heart syndrome, in three generations: *short report*, 328
Intracranial teratoma, giant, associated with pulmonary hypoplasia, prenatal diagnosis: *brief paper*, 880
Intrauterine growth retardation, severe, blepharophimosis, and cylindrical nose with midline groove, new syndrome? *short report*, 525
Iris coloboma, ptosis, hypertelorism, and mental retardation: Baraitser-Winter syndrome or Noonan syndrome? *short report*, 425
Italian infants with limb/pelvis-hypoplasia/aplasia syndrome, confirmation of autosomal recessive inheritance: *short report*, 65: *letter*, 797

Jacobs, Professor Pat, standing down as Cytogenetics editor, 1043
Jarcho-Levin syndrome, airway abnormalities, two cases: *brief report*, 875
Journal abstracts, 85, 174, 261, 349, 443, 532, 620, 707, 796, 885, 972

Kallmann syndrome, and X linked ocular albinism, genetic mapping of gene loci, 923
Klinefelter's syndrome, with probable de novo 17q duplication: *short report*, 436

LADD syndrome and split hand/split foot deformity in a family, overlap between the EEC and LADD syndromes: *brief paper*, 700
Lambdoid synostosis, and humeroradioulnar synostosis: *short report*, 81
Leprechaunism, insulin resistance resembling, 5 sibs, 470
Leukaemogenesis, molecular aspects of P53 in lymphoma, 339 *abs*
Li-Fraumeni syndrome, possible variant, 335 *abs*
Limb reduction defects, ear anomalies, and clefting, new autosomal recessive condition? 342 *abs*
Limb/pelvis-hypoplasia/aplasia syndrome
Bedouin patient and review: *short reporet*, 62: *letter*, 797
two Italian sibs and further confirmation of autosomal recessive inheritance: *short report*, 65: *letter*, 797
Linkage disequilibrium between Huntington's disease locus and markers at loci D4S10, D4S95, and D4S111, Northern Ireland, 1018
Linkage studies versus association in psychosis genetics: *annotation*, 634; *comment*, 638
Low density lipoprotein receptor gene, six DNA polymorphisms, 273
Low segregation ratios in autosomal recessive disorders: *annotation*, 449
Lymphoma, and leukaemogenesis, molecular aspects of P53, 339 *abs*
Lysosomal enzyme, molecular analysis in Gaucher's disease: *review article*, 889

Magnetic Activated Cell Sorter, for prenatal diagnosis from maternal blood, 1051
Malformed man, elderly, chromosome 20 long arm deletion: *short report*, 171
Malignant hyperthermia, genetics: *review article*, 89
Mandibular prognathism, genetics, analysis of large European noble families, 112
Marfan syndrome, screening of fibrillin gene FBN1 for causal mutations, 348 *abs*
Marshall-Smith syndrome, without respiratory complications, long survival: *brief paper*, 877
Martin Bell family, link with fragile X syndrome, 341 *abs*
Maternal blood, prenatal diagnosis from, Magnetic Activated Cell Sorter, 1051
Medical genetics, advances in brief, 85, 174, 261, 349, 443, 532, 620, 707, 796, 885, 972
Medium chain acyl-CoA dehydrogenase deficiency, screening for G 985 mutation, Scottish women, 346 *abs*
Medulloblastoma, congenital anomalies, and Gorlin syndrome, 337 *abs*
Mendelian cytogenetics, chromosome rearrangements associated with mendelian disorders: *review article*, 713
Menkes disease, cytogenetic survey, implications for detection of chromosomal rearrangements in X linked disorders: *short report*, 314
Menkes syndrome, cloning of candidate gene spanning MNK translocation breakpoint, 334 *abs*
Mental retardation
deafness, or aphasia: *letter*, 262
deafness, and familial ataxia, new form: *brief paper*, 694
iris coloboma, ptosis, and hypertelorism: Baraitser-Winter syndrome or Noonan syndrome? *short report*, 425
profound, characteristic facies, premature balding, muscular hypotrophy and small patellae, two unrelated male patients: *short report*, 319
X linked, and *FRAXE* fragile site, 97
Meryon, Edward (1809-1880), and muscular dystrophy: *portraits in medical genetics*, 506
Methylation, parental allele specific, of IGF-II, and Beckwith-Wiedemann syndrome, 353
Microcephaly, seizures, mental retardation, CHD, and skeletal abnormalities, continuation of a case report: *letter*, 445
Microphthalmos, classification, 664

Microsatellite repeats, visualisation by silver staining, 346 *abs*
Mitochondrial cardiomyopathy, X linked congenital, possible, three families, 269
Mixoploidy, 46,XX/69,XXX, resulting from second polar body incorporation into blastomere: *short report*, 597
Molecular diagnosis of some common genetic diseases, Russia and former USSR: *medical genetics around the world*, 141
Molecular genetic diagnosis, evaluation in management of familial adenomatous polyposis coli, 675
Monogenic disorders, analysis of a terminal Xp22.3 deletion, 838
Mosaicism for duplication 12q (12q13–q24.2) in dysmorphic male infant: *short report*, 70
MR/MCA syndrome, with triple structural mosaicism of chromosome 18 and abnormal skin pigmentation: *short report*, 614
Multiple sclerosis, wrongly diagnosed for X linked adrenomyeloneuropathy, 338 *abs*
Muscular dystrophy
  and Edward Meryon (1809-1880): *portraits in medical genetics*, 506
  facial involvement absence may differentiate a scapular onset from from FSH dystrophy, 338 *abs*
  limb-girdle, autosomal recessive adult forms, genetic heterogeneity, Brazil, 385
  rapid analysis of dystrophin gene deletions shows variable electrophoretic mobility, 60
  Xp21, variability of clinical, genetic, and protein abnormalities in manifesting carriers, 337 *abs*
  Xp21 linked, integrated study of 100 patients. Part 1. Trends across clinical groups, 728: Part 2. Correlations within individual patients, 737: Part 3. Differential diagnosis and prognosis, 745
Myelomeningocele, and Waardenburg syndrome, in a family: *short report*, 83
Myopathy, congenital, with fibre type disproportion, 348 *abs*
Myotonia, autosomal recessive generalised (Becker), evidence for genetic homogeneity, 914
Myotonic dystrophy
  DNA methylation study, 189
  molecular diagnosis: *letter*, 533
  specific mutation: *review article*, 978
  unstable CTG repeat, 341 *abs*
  in various tissues from fetuses predicted to be at high risk, analysis of expansion associated with, 341 *abs*

Naevoid basal cell carcinoma syndrome, complications, 460
Nager acrofacial dysostosis: *syndrome of the month*, 779
Nasal cilia, immotile, and bronchiectasis, associated with Usher syndrome type I, two brothers: *short report*, 253
Neural tube defect
  and chromosome abnormalities, biochemical screening, 336 *abs*
  familial cases, combinations of sexes: *letter*, 447
  lesion descriptions made by different European pathologists, 942
  upper and lower, alternate hypothesis, 849
Neurofibromatosis clinic, Oxford, first two years' experience, 3376 *abs*
Neurofibromatosis type 1, knowledge, experience, and reproductive decisions of affected patients and families, 567
Neurofibromatosis type 1, genetic study, PCR based approach, Ontario, 363
Noonan syndrome
  or Baraitser-Winter syndrome: iris coloboma, ptosis, hypertelorism, and mental retardation: *short report*, 425
  male fertility in, 342 *abs*
Norrie's disease, manifesting heterozygosity, 346 *abs*
Northern Ireland
  identification of CAG repeat in Huntington's disease gene (IT15) in benign hereditary chorea, 1012
  significant linkage disequilibrium between Huntington's disease locus and markers at loci D4S10, D4S95, and D4S111, 1018
Notices, 88, 536, 624, 800, 974
Nystagmus, congenital, cosegregating with balanced 7;15 translocation: *short report*, 526

Ocular albinism, X linked, and Kallmann syndrome, genetic mapping of gene loci, 923
Orofaciodigital syndrome type III in two sibs: *brief paper*, 870
Osteoarthritis, COL2A1, molecular analysis, 348 *abs*
Osteogenesis imperfecta
  'lethal', survival, 337 *abs*
  mutation detection in gene encoding alpha1 chain of type I collagen, 335 *abs*
  recurrent severe, molecular and genetic mechanisms, 339 *abs*
Osteogenesis imperfecta type III, mutations in type I collagen structural genes, COL1A1 and COL1A2, are not necessarily responsible, 492
Otopalatodigital syndrome type II: *syndrome of the month*, 310
Ovarian cancer
  material, fresh and archival, p53 tumour suppressor gene mutations, 335 *abs*
  p53 mutations, 339 *abs*
  psychosocial issues raised by familial register, 575
Oxford neurofibromatosis clinic, first two years' experience, 3376 *abs*

P53
  in lymphoma and leukaemogenesis, molecular aspects, 339 *abs*
  mutations, in ovarian cancer, 339 *abs*
Patellae, small, profound mental retardation, characteristic facies, premature balding and muscular hypotrophy, two unrelated male patients: *short report*, 319
Patent ductus arteriosus and unusual face in large family, 503
PEHO syndrome, diagnostic criteria and genetics, 932
Pentalogy of Fallot, acrocephalopolysyndactyly, and hypoacusis, with 46,XX,t(1;18)(p31;q11): *short report*, 438
Perineal hypospadias, androgen receptor gene mutations identified, 198
Phenylketonuria
  genetic background of clinical homogeneity, Poland, 232
  molecular analysis of contiguous exons of phenylalanine hydroxylase gene, 228
  R261Q mutation and maternal PKU, 284
  rapid and efficient PCR/*Sty*I test for identification of common mutation R408W: *technical note*, 153
  Turkish patients, mutation analysis, 129
  untreated, comparison of genotype and intellectual phenotype, 401
Pituitary abnormalities and anophthalmia, and deletion 14(q22.1→q22.3): *short report*, 251
Poland anomaly with contralateral ulnar ray defect: *short report*, 423
Polycystic kidney disease
  autosomal dominant
    childhood onset, in sibs, clinical picture and recurrence risk, 583
    estimating locus heterogeneity, Spanish population, 910
Polycystic kidneys, bilateral, familial, with skeletal malformations, molecular genetic diagnosis: *short report*, 419: *letter*, 973
Polydactyly, five generation Indian family, 296
Polymerase chain reaction
  diagnosis of infantile spinal muscular atrophy using amniocyte and CVS DNA: *short report*, 162
  single cell, reliability, 332 *abs*
  /*Sty*I test, rapid and efficient, for identification of common mutation R408W in phenylketonuria patients: *technical note*, 153

technique, and forensic medicine: *review article*, 625
Prader-Willi-like phenotype, in fragile X patients, clinical and molecular studies, 761
Prader-Willi syndrome
or Angelman syndrome, uniparental disomy, in patients with an additional small inv dup(15) chromosome, 756
molecular genetic studies, 348 *abs*
Pregnancy, second trimester termination, reproductive behaviour after, 344 *abs*
Prenatal diagnosis
of diastrophic dysplasia with polymorphic DNA markers, 265
from maternal blood, Magnetic Activated Cell Sorter, 1051
of giant intracranial teratoma associated with pulmonary hypoplasia: *brief paper*, 880
of a hypermethylated full fragile X mutation in chorionic villi of a male fetus: *brief paper*, 785
Pseudomosaicism, apparent, at amniocentesis, and hypomelanosis of Ito associated with mosaicism for trisomy 7: *brief paper*, 783
Pseudotrisomy 13 and autosomal recessive holoprosencephaly: *short report*, 970
Psychosis genetics, association versus linkage studies: *annotation*, 634; *comment*, 638
Psychosocial genetics
DMD carrier risk, women's understanding, 562
effects of prenatal screening for cystic fibrosis, 543
emerging scientific discipline: *editorial*, 537
Huntington's disease testing
demand, attitudes, and psychological assessment, 549
perception by young women, preferring uncertainty to certainty?, 557
issues raised by familial ovarian cancer register, 575
neurofibromatosis type 1, knowledge, experience, and reproductive decisions of affected patients and families, 567
Ptosis, iris coloboma, hypertelorism, and mental retardation: Baraitser-Winter syndrome or Noonan syndrome? *short report*, 425
Pulmonary hypoplasia, and giant intracranial teratoma, prenatal diagnosis: *brief paper*, 880
Pulmonary vascular occlusive disease, with other anomalies, continuation of a case report: *letter*, 445
Pyloric stenosis, linkage analysis and markers from chromosome 9q11-q33, 393

Restriction fragment length polymorphism
analysis for APP 717 mutations associated with Alzheimer's disease, 476
new, at DXS101 locus, allows carrier detection in family with X linked agammaglobulinaemia: *short report*, 512
Retinitis pigmentosa
characterisation of a 340 kb YAC and cDNA enrichment, 335 *abs*
Scottish population, mutation detection in rhodopsin and peripherin genes, 347 *abs*
X linked, genetic localisation of RP2 type, 345 *abs*
Retinoblastoma, possible link with low level radiation: *conference report*, 440
Retinoschisis disease gene, and characterisation of highly polymorphic microsatellite at *DXS207* locus, 300
Ring chromosome 18, transmission from mother with 46,XX/47,XX,+r(18) mosaicism to daughter, resulting in 46,XX,r(18) karyotype: *brief paper*, 964
Ring (X) chromsomes and a severe phenotype, three patients, 482
Romano-Ward syndrome
counselling pitfalls, consanguinity, cardiac arrest, hearing impairment, and ECG abnormalities: *short report*, 325: *letter*, 886
evidence of genetic and phenotypic heterogeneity, 947
Russia and former USSR, molecular diagnosis of some common genetic diseases: *medical genetics around the world*, 141

S549N mutation in Pakistani family, absence of cystic fibrosis mutations in Asian population: *short report*, 164
Sacral agenesis and holoprosencephaly in fetus with terminal deletion 7q36→7qter: *short report*, 521
Schinzel, Professor Albert, taking over as Cytogenetics editor, 1043
Schizophrenia
cosegregation with Becker muscular dystrophy, 131
and homozygosity, association between, at dopamine D3 receptor gene: *letter*, 708
Scleroatrophic syndrome of Huriez, 342 *abs*
Scotland
CAG repeat in Huntington's disease and age of onset, 1014
Huntington's disease associated trinucleotide repeat, 1003
Sex, diagnosis
preimplantation, in X linked disease, 332 *abs*
prenatal, from maternal peripheral blood using PCR, 336 *abs*
Short rib-polydactyly syndromes, lethal, further evidence for their overlapping in a continuous spectrum, 937
Skeletal dysplasia, lethal, with generalised sclerosis and advanced skeletal maturation, Blomstrand chondrodysplasia? *short report*, 155
Skewed X inactivation, is it responsible for symptoms in female carriers for adrenoleucodystrophy?, 651
Social and ethical issues in presymptomatic testing for Huntington's disease, European Community collaborative study, 1028
Sotos syndrome, search for uniparental disomy, 342 *abs*
Sparse hair, short limbed dwarfism, genital hypoplasia, and vertebral anomalies: Ellis-van Creveld syndrome variant? *short report*, 322
Spastic paraplegia, X linked, clinical heterogeneity at a single gene locus, 381
Specialty of clinical genetics, European Society of Human Genetics survey, 147
Spina bifida
lesion, location, sex differences: *letter*, 262
number of births, trends, 338 *abs*
Spinal muscular atrophy, infantile, rapid diagnosis by direct amplification of amniocyte and CVS DNA: *short report*, 162
Spinobulbar muscular atrophy, specific mutation: *review article*, 978
Spinocerebellar ataxia type 1, autosomal dominant, presymptomatic testing: *short report*, 616
Spinocerebellar atrophy, specific mutation: *review article*, 978
Split hand/foot, isolated, Hungary, 1975-1984, epidemiological study, 593
Split hand/split foot deformity and LADD syndrome in a family, overlap between the EEC and LADD syndromes: *brief paper*, 700
Spondyloepiphyseal dysplasia congenita, resulting from substitution of glycine 997 by serine in alpha-1(II) chain of type II collagen, clinical features, 27
SRY gene, new de novo mutation (A113T) in high mobility group box leads to XY gonadal dysgenesis, 655
Strabismus, delayed speech development, facial asymmetry, and transverse ear lobe creases, new syndrome: *short report*, 76
Strawberry naevi, multiple, and severe developmental delay, a new syndrome? *short report*, 883
Suicide risk, in Huntington's disease, 293

Tay-Sachs disease
HEXA gene intron 9 donor splice site mutation frequently found in non-Jewish patients from the British Isles, 479

late infantile, two new mutations are both in exon 1 of beta-hexosaminidase alpha subunit gene, 123
Tetrahydrobiopterin deficiency, dihydropteridine reductase gene, two new mutations, 465
Thalassaemia, Filipino β°, high Hb A_2 β° thalassaemia resulting from large deletion of 5′ globin gene region, 240
Thalassaemia-beta heterozygotes in Brazil, molecular characterisation: *letter*, 797
Thrombocytopenia, X linked recessive: *short report*, 968
Translocation
balanced 7;15, congenital nystagmus cosegregating: *short report*, 526
balanced, (14), reproductive possibilities for carriers in families with partial trisomy of proximal 14q: *short report*, 73
reciprocal, balanced whole arm, resulting in three different adverse pregnancy outcomes: *short report*, 417
t(15;20)(q13;q13.1), chiasma frequency, quadrivalent orientation, and first meiotic segregation, 336 *abs*
unbalanced, in Waardenburg gene region, two sibs: *short report*, 607
Transthyretin variant (alanine 49) associated with familial amyloidotic polyneuropathy, French family, 117
Transthyretin variant (alanine 71) associated with familial amyloidotic polyneuropathy, French family, 120
Transverse ear lobe creases, delayed speech development, facial asymmetry, and strabismus, new syndrome: *short report*, 76
Triple structural mosaicism of chromosome 18, in child with MR/MCA syndrome and abnormal skin pigmentation: *short report*, 614
Trisomy
partial, of proximal 14q, reproductive possibilities for balanced translocation (14) carriers: *short report*, 73
7, hypomelanosis of Ito with mosaicism for, and apparent 'pseudomosaicism' at amniocentesis: *brief paper*, 783
9 mosaicism, in two girls with multiple congenital malformations and mental retardation: *short report*, 433
10qter, confirmed by in situ hybridisation: *short report*, 601
18, rapid confirmation using directly fluoresceinated 18 centromere specific probe, 336 *abs*
19q (q13.2→qter), owing to unusual maternal translocation: *brief paper*, 697
Tuberous sclerosis
identification of markers flanking locus on chromosome 9, 224
variability of expression, 41
Turkish phenylketonuria patients, mutation analysis, 129
Turner stigmata on Yp, chromosomal localisation of gene(s) for, 918
Turner's syndrome
detection by MSAFP/Free beta (hCG) screening, 340 *abs*
screening for occult Y mosaicism, 346 *abs*

Ulnar ray defect, contralateral, and Poland anomaly: *short report*, 423
Usher syndrome type I, associated with bronchiectasis and immotile nasal cilia, two brothers: *short report*, 253
Usher syndrome type II
audiometric identification of carriers, 343 *abs*
genetic heterogeneity, 843

Valvular dysplasia, sex linked: *brief paper*, 873
Vas deferens, congenital absence, and CF carrier frequency: *letter*, 973
Velocardiofacial syndrome
and 22q11 microdeletions: *editorial*, 801, 807
22q11 microdeletions, prevalence, implications for genetic counselling and prenatal diagnosis, 813
in mother and daughter, variability of clinical phenotype, 825
new marker and conserved sequences close to, 818
Vertebral anomalies, short limbed dwarfism, genital hypoplasia, and sparse hair: Ellis-van Creveld syndrome variant? *short report*, 322
von Hippel-Lindau disease
on chromosome 3p25-26, dinucleotide repeat polymorphisms, genetic mapping, 487
neoplasms, five types, common mechanism of tumourigenesis, 339 *abs*
tumour suppressor gene, detailed genetic mapping, 104

Waardenburg gene region, unbalanced translocations, two sibs: *short report*, 607
Waardenburg syndrome, and myelomeningocele, in a family: *short report*, 83
Weyer's ulnar ray/oligodactyly syndrome: *letter*, 533
Williams-Beuren syndrome, and chromosome abnormalities: *letter*, 886
Wilms's tumour gene, WT1, mutational screening, in males with genital abnormalities, 767
Wolf-Hirschhorn syndrome, paternal origin of submicroscopic deletion of chromosome 4, 348 *abs*

X chromosome inactivation, and X linked disease diagnosis, females, 177
X linked adrenomyeloneuropathy, misdiagnosed as multiple sclerosis, 338 *abs*
X linked agammaglobulinaemia
isolation of defective gene: *annotation*, 452
new RFLP at DXS101 locus allows carrier detection: *short report*, 512
X linked congenital mitochondrial cardiomyopathy, possible, three families, 269
X linked form of hyper IgM syndrome (HIGM1), mapping, 202
X linked hydrocephalus, refining genetic location of gene within Xq28, 214
X linked mental retardation
and *FRAXE* fragile site, 97
non-specific, gene mapping, evidence for linkage to chromosomal region Xp21.2–Xp22.3: *brief paper*, 866
X linked spastic paraplegia (SPG2), clinical heterogeneity at a single gene locus, 381

Young-Simpson syndrome, parental consanguinity: *short report*, 255
Yunis-Varón syndrome, and congenital heart malformation: *brief paper*, 788